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## Annual report, 2003-2004

Jouni Uitto Thomas Jefferson University

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# DEPARTMENT OF DERMATOLOGY AND CUTANEOUS BIOLOGY Jefferson Medical College

# **ANNUAL REPORT 2003-2004**

Jouni Uitto, MD, PhD Professor and Chair



## DEPARTMENT OF DERMATOLOGY AND CUTANEOUS BIOLOGY JEFFERSON MEDICAL COLLEGE

#### STATEMENT OF THE CHAIR Academic Year 2003-2004

Jouni Uitto, M.D., Ph.D. August 15, 2004

#### **PREFACE**

This Academic year marks the 18<sup>th</sup> year of my tenure as the Chair of the Department of Dermatology and Cutaneous Biology at Jefferson Medical College. Over the past two decades, our Department has witnessed extraordinary growth in the academic enterprise. In fact, at the end of the Academic Year 2003, we currently have a full-time faculty of 24, comprising of 10 clinicians and 14 research faculty, the total number of trainees currently stands at 21, including 12 residents and 9 research fellows, our clinical enterprise continues to flourish, and extramural grant support continues to grow.

At the same time, the Department of Dermatology and Cutaneous Biology, like many other departments at Jefferson, continue to face challenges of increasing costs and expenses, while cutbacks in the guise of reduced reimbursements, soaring of malpractice costs, and reductions in the institutional support for growth and maintenance of the departmental infrastructure, have posed major challenges. In spite of these obstacles, our department is proud of its standing as one of the leading institutions nationwide in providing quality dermatologic care and performing state-of-the-art cutaneous biology research.

I am pleased to present this report on the clinical, teaching, and research activities of the Department of Dermatology and Cutaneous Biology at Jefferson Medical College during the academic year 2003-2004.

#### **CLINICAL ACTIVITIES**

Our clinical enterprise, centered on our on-campus practice location at 833 Chestnut Street, has experienced continued growth. Under the direction of Young C. Kauh, MD, Professor and Senior Vice Chair of Departmental Development, and Guy F. Webster, MD, Ph.D., Professor and Clinical Vice Chair, we have reached a new record of patient visits during this academic year. Contributing to our clinical success have been other clinicians, including Dr. Tanya Humphreys, Director of Cutaneous Surgery, Dr. Jason Lee, Director of Dermatopathology, Dr. Patrice Hyde, Chief of Pediatric Dermatology, and Franziska Ringpfeil, Assistant Professor of Dermatology. During this academic year, four clinicians have left full-time employment of the Department: Dr. Paul Bujanauskas joined the Clinical Faculty of Johns Hopkins University; Dr. Magaly DelMonaco entered into private practice in New Jersey; Dr. Norman B. Sykes entered into private practice in Ellsworths, Maine. The departure of these physicians coincides with the departmental decision to close, based on prudent fiscal analysis, our off-site independent satellite

practices. In addition, Dr. Kehua Li is undertaking his dermatopathology fellowship at the University of Colorado. We thank them for their long-term contributions to the Department's clinical activities. At the same time, we have been successful in recruiting two additional new clinicians: Dr. Stacy Katchman and Dr. Barbara Vail have joined the Department as members of the full-time faculty, and we welcome them to Jefferson Dermatology.

Mr. Alan Strigga, the Departmental Business Manager, with the help of Karen Griffin, Clinical Manager, has been instrumental in maintaining the high quality of standards of our clinical operations. We thank them, and the entire clinical office staff, for their superb help.

#### **Specialty Services**

In addition to general dermatology, a number of specialty services are available to our patients at our Center City offices.

<u>The Cutaneous Surgery Unit</u>, headed by Dr. Tanya Humphreys, offers a state-of-the-art array of surgical services, including Mohs micrographic surgery, laser surgery, and a number of cosmetic procedures.

<u>The Division of Pediatric Dermatology</u>, is directed by Dr. Patrice Hyde who, in addition to our center city office, practices at A.I. DuPont Hospital for Children. Over the past several years, we have also expanded our pediatric dermatology services to Temple Children's Hospital under the direction of Dr. Franziska Ringpfeil, Assistant Professor of Dermatology.

#### **Molecular Diagnostics**

The clinical services are augmented by Molecular Diagnostics of Heritable Skin Diseases Clinic attended by myself and Dr. Gabriele Richard, Associate Professor of Dermatology and Cutaneous Biology, a dermatologist, genetic counselor, and molecular geneticist, with extensive expertise and experience in clinical and molecular genetic studies of ichthyosis and other disorders of cornification. Dr. Richard's research has elucidated the molecular basis of numerous ichthyoses, palmar plantar keratodermas and erthyrokeradermias, and she is a recognized expert in research on connexins disorders. Dr. Richard also serves with me as the codirector of the Molecular Diagnostics Laboratory located in our Department.

#### **Dermatopathology:**

The dermatopathology service of the Department of Dermatology and Cutaneous Biology is directed by Dr. Jason Lee, Clinical Assistant Professor of Dermatology and Cutaneous Biology. Since joining us in 2002, this service has grown to – accessions per year, which provides an excellent source of teaching material for our residency training program. We have realigned our dermatopathology teaching with complementary services within the Department of Pathology that is responsible for hospital-based diagnostics. We expect our dermatopathology services to grow, and we look forward a potential recruitment of an additional dermatopathologist in the near future.

In summary, the clinical enterprise of the Department of Dermatology and Cutaneous Biology continues to offer broad and diversified specialty services to our patients, and we look forward to continued expansion of these services. The clinical faculty deserves my congratulations for their outstanding work.

#### **TEACHING**

#### **Residency Training Program**

Our Residency Training Program continues to be fully accredited towards the eligibility for certification by the American Board of Dermatology. The program currently has twelve residents in good standing. In June 2004, three of our residents completed their training: The Chief Resident, Dr. Judith Cenci, entered private practice in Sellersville, PA; Dr. John Mulholland entered a dermatopathology fellowship program at University of California, Irvine; Dr. Subhadra Sundaram, entered a cutaneous surgery fellowship at Harvard University . I would also like to extend our thanks to our current chief resident, Dr. Christine Ambro, for doing an outstanding job.

#### **Medical Student Teaching**

The Department participates in the medical student training by giving – lectures in dermatology for the second year students. These lectures have been extremely well received by the students as judged from the evaluation feedback. In addition, the Department offers dermatology electives to the medical students, primarily from Jefferson Medical College, but space permitting, also from other universities. Dr. Jason Lee has assumed a leadership role by serving as a departmental liaison in coordinating the medical students' educational activities. All the clinical work by the students is under direct supervision by the full-time clinical faculty.

In addition to medical students, our clinics have a number of rotators, including House Staff members from other Jefferson Departments, as well as from other U.S. medical schools and from abroad. During this academic year, we have had international clinical fellows from Brazil, Korea, Pakistan, and Spain, among others.

#### RESEARCH Research Faculty

Our research faculty has remained stable and has continued to be a successful in biomedical research. During the past year, several promotions in the academic rank of the research faculty took place. Dr. Kyonggeun Yoon was promoted to the rank of Professor and Dr. Olga Igoucheva to Research Assistant Professor.

Congratulations for these well-deserved promotions!

The extramural support to our research has remained stable, and the Department of Dermatology and Cutaneous Biology has continued to successfully compete for grant support from the National Institutes of Health and other sources. In fact, the Department has remained among the top five dermatology departments in the country in NIH ranking, having been no. 1 in 1998. In addition, Dr. Andrzej Fertala continues to receive grant support from NASA while Drs. My

Mahoney, John Klement and Uli Rodeck have funding from the Department of Defense. Drs. Vitali Alexeev, Reza Ghohestani, Mỹ Mahoney, Fatima Rouan, Gabriele Richard and Franziska Ringpfeil have been recipients of Career Development Awards from a variety of sources, including the National Institutes of Health, Dermatology Foundation, and the American Skin Association. While these Career Development Awards are reaching their end, the challenge is to obtain steady funding that will guarantee continued support to the research of these young investigators. This challenge is compounded by increasing competition for federal support to biomedical research. Finally, I would like to express our thanks to Dr. Mon-Li Chu, Professor and Vice Chair of Research, for her contributions in support of further development of our departmental research enterprise.

#### **Jefferson Institute of Molecular Medicine**

The Institute continues to serve as an umbrella for broad inter-departmental and inter-disciplinary research at Jefferson with emphasis on matrix biology and pathology, cancer biology and human molecular genetics. The Institute provides core facilities that serve not only the Departmental researchers but also the entire Jefferson research community at large. The current cores include: Tissue culture core (Director: Dr. Ellen Pfendner); Morphology core (Director: ?; shared with the Department of Pathology); Transgenic animal facilities (Director: Dr. John F. Klement); and DNA syntheses and sequencing core (Co-Directors: Dr. Mon-Li Chu and Dr. Hanus Alder; shared with the Kimmel Cancer Center). Other functions of the Institute include organization of on-going seminar and science lecture series in coordination with Orthopedic Research and Rheumatology/Medicine as well as departmental research in progress series.

#### **DebRA Molecular Diagnostics Laboratory**

This laboratory was established in 1996 as a global referral center for molecular diagnostics of epidermolysis bullosa (EB), as group of heritable blistering disorders. This laboratory, a partnership between the lay organization, the Dystrophic Epidermolysis Bullosa Research Association of America, and the Department of Dermatology and Cutaneous Biology at Jefferson Medical College, has continued to provide DNA diagnostics to the global network of EB patients. In fact, as of today, the Center has analyzed DNA from over a thousand families with different forms of EB and has performed close to 200 prenatal diagnoses in these families. Dr. Ellen Pfendner, Assistant Professor of Dermatology, has served as the Manager of this increasingly busy laboratory. The laboratory has expanded its services to other heritable skin diseases, including various keratinization disorders; this effort has been spearheaded by Dr. Gabriele Richard, Co-Director of the laboratory. Over the past year, the laboratory has also partnered with PXE International, a patient advocacy organization supporting research on the causes and treatment of pseudoxanthoma elasticum, a group of disorders characterized by calcification of elastic structures in the skin, eyes, and cardiovascular system. As a result of this partnership, the laboratory has identified disease-causing mutations in a cohort of ~200 families with PXE and has participated in development of a diagnostic kit for these mutations.

#### JEFFERSON CENTER FOR INTERNATIONAL DERMATOLOGY

Over the past two decades, Jefferson Dermatology has become increasingly recognized as a leader in international dermatology. These activities are concentrated under the umbrella of Jefferson Center for International Dermatology which was established in 1987 to promote the exchange of information and dermatological knowledge within the global network of international dermatology. Within this framework, Jefferson Dermatology ahs continued to host visiting residents, scholars, and scientists from a variety of countries. Finally, the Center continues to offer a Certificate Program which was developed to provide physicians from abroad the opportunity to learn the most current therapies available and to observe the new skills and techniques being used in the field of dermatology.

#### **Summary**

The Department of Dermatology and Cutaneous Biology at Jefferson Medical College continues to excel in academic enterprise, including clinical activities, research endeavors and teaching programs. The quality of our research is attested to by our high ranking as one of the leaders in research in cutaneous biology and pathology. Our international relations continue to flourish and bring recognition not only to the Department but also to Jefferson Medical College at large. The Department is fully committed to further development of activities in the areas of research, education and patient care, to the maximum extent allowed by fiscal realities.

On behalf of the entire Department of Dermatology and Cutaneous Biology at Jefferson, I would like to thank the President and the Board of Trustees of Thomas Jefferson University, the Dean of Jefferson Medical College, and the President of Thomas Jefferson University Hospital, for their continued support to the programs of the Department of Dermatology and Cutaneous Biology.

Jouni Uitto, MD, PhD
Professor and Chair
Department of Dermatology and Cutaneous Biology
Director, Jefferson Institute of Molecular Medicine

#### **FACULTY AND TRAINEES**

#### <u>DEPARTMENT OF DERMATOLOGY AND CUTANEOUS BIOLOGY</u> 2003 - 2004

#### **CHAIRMAN**

**Jouni Uitto, MD, PhD**, Professor of Dermatology and Cutaneous Biology; Professor of Biochemistry and Molecular Pharmacology (secondary appointment); Director, Jefferson Institute of Molecular Medicine

#### **VICE CHAIRS**

- **Young C. Kauh, MD**, Professor and Senior Vice Chairman of Departmental Development, Department of Dermatology and Cutaneous Biology
- **Guy F. Webster, MD, PhD**, Professor and Clinical Vice Chair, Department of Dermatology and Cutaneous Biology; Professor of Medicine, Division of Clinical Pharmacology (secondary appointment); Director, Center for Cutaneous Pharmacology, Department of Dermatology and Cutaneous Biology
- **Mon-Li Chu, PhD,** Professor and Vice Chair of Research, Department of Dermatology and Cutaneous Biology; Professor of Biochemistry and Molecular Pharmacology (secondary appointment).

#### **DIVISION CHIEFS/DIRECTORS**

- **Tatyana R. Humphreys, MD**, Clinical Associate Professor of Dermatology and Cutaneous Biology; Director, Center for Laser Surgery and Cosmetic Dermatology
- **Patrice M. Hyde, MD**, Clinical Assistant Professor of Dermatology and Cutaneous Biology; Chief of Pediatric Dermatology
- **Jason B. Lee, MD,** Assistant Professor of Dermatology and Cutaneous Biology; Director, Dermatopathology of the Department of Dermatology and Cutaneous Biology

#### **HONORARY MEMBERS:**

- **Joseph K. Corson, MD,** Honorary Clinical Associate Professor of Dermatology and Cutaneous Biology
- **Charles H. Greenbaum, MD**, Honorary Clinical Professor of Dermatology and Cutaneous Biology
- **Richard H. Musgnug, MD**, Honorary Assistant Professor of Dermatology and Cutaneous Biology

#### **PROFESSORS**

Mohamed A. Amer, MD, Visiting Professor of Dermatology and Cutaneous Biology
 Mon-Li Chu, PhD\*, Professor of Dermatology and Cutaneous Biology
 Isao Hashimoto, MD, PhD, Visiting Professor of Dermatology and Cutaneous Biology
 Lasse O. Kanerva, MD, PhD, Visiting Research Professor of Dermatology and Cutaneous Biology
 Cutaneous Biology

Francisco Kerdel-Vegas, MD, Visiting Professor of Dermatology and Cutaneous Biology
Kari I. Kivirikko, MD, PhD, Visiting Professor of Dermatology and Cutaneous Biology
Torello Lotti, MD, Visiting Professor of Dermatology and Cutaneous Biology
Henry C. Maguire, MD, Research Professor of Dermatology and Cutaneous Biology
Thomas O'Brien, PhD, Professor of Dermatology and Cutaneous Biology
Joseph L. Pace, MD, Visiting Professor of Dermatology and Cutaneous Biology
Lawrence C. Parish, MD, Clinical Professor of Dermatology and Cutaneous Biology
Yoon-Kee Park, MD, PhD, Visiting Professor of Dermatology and Cutaneous Biology
Ulrich Rodeck\*, MD, Professor of Dermatology and Cutaneous Biology
Terence J. Ryan, FRCP, Visiting Professor of Dermatology and Cutaneous Biology
Markku Ryynänen, MD, PhD, Visiting Professor of Dermatology and Cutaneous Biology
Jouni Uitto, MD, PhD\*, Professor of Dermatology and Cutaneous Biology, Professor of Biochemistry and Molecular Pharmacology

**Guy F. Webster, MD, PhD\***, Professor of Dermatology and Cutaneous Biology, Professor of Medicine, Division of Clinical Pharmacology

#### **ASSOCIATE PROFESSORS**

Sirpa Aho, PhD\*, Research Associate Professor of Dermatology and Cutaneous BiologyGuy F. Carnabucci, MD, Adjunct Clinical Associate Professor of Dermatology and Cutaneous Biology

Andrzej Fertala, PhD\*, Associate Professor of Dermatology and Cutaneous Biology
 David J. Herrick, PhD\*, Research Associate Professor of Dermatology and Cutaneous Biology

**Tatyana R. Humphreys, MD\***, Clinical Associate Professor of Dermatology and Cutaneous Biology, Chief, Division of Cutaneous Surgery, Director, Center for Laser Surgery and Cosmetic Dermatology

Jaakko Karvonen, MD, PhD, Visiting Professor of Dermatology and Cutaneous Biology Bernard A. Kirshbaum, MD, Adjunct Clinical Associate Professor of Dermatology and Cutaneous Biology

**Robert G. Knowlton, PhD**, Adjunct Associate Professor of Dermatology and Cutaneous Biology

Mỹ Mahoney, PhD\*, Associate Professor of Dermatology and Cutaneous Biology Alain Mauviel, PhD, Adjunct Associate Professor of Dermatology and Cutaneous Biology<sup>12</sup>

**Leena Pulkkinen, PhD**, Adjunct Research Associate Professor of Dermatology and Cutaneous Biology

- Gabriele Richard, MD\*, Associate Professor of Dermatology and Cutaneous Biology Janet Sawicki, PhD, Research Associate Professor of Dermatology and Cutaneous Biology
- **Kyonggeun Yoon, PhD\***, Associate Professor of Dermatology and Cutaneous Biology, Director, Cutaneous Gene Therapy Center
- **John A. Zitelli, MD**, Adjunct Clinical Associate Professor of Dermatology and Cutaneous Biology

#### **ASSISTANT PROFESSORS:**

- Vitali Alexeev, PhD\*, Assistant Professor of Dermatology and Cutaneous Biology Ercem Atillasoy, MD, Adjunct Clinical Assistant Professor of Dermatology and Cutaneous Biology
- **Harold L. Colburn, Jr., MD**, Clinical Assistant Professor of Dermatology and Cutaneous Biology
- **Chalmers E. Cornelius, III, MD**, Assistant Professor of Dermatology and Cutaneous Biology
- **Magaly del Monaco, DO,** Adjunct Clinical Assistant Professor of Dermatology and Cutaneous Biology
- **Reza F. Ghohestani, MD, PhD\***, Research Assistant Professor of Dermatology and Cutaneous Biology
- **Seung K. Hann, MD, PhD**, Visiting Clinical Assistant Professor of Dermatology and Cutaneous Biology<sup>13</sup>
- **Patrice Hyde, MD\*,** Clinical Assistant Professor of Dermatology and Cutaneous Biology, Chief, Pediatric Dermatology
- **Stacy D. Katchman, MD\***, Clinical Assistant Professor of Dermatology and Cutaneous Biology
- John F. Klement, PhD\*, Assistant Professor of Dermatology and Cutaneous Biology Raymond Krain, MD, Clinical Assistant Professor of Dermatology and Cutaneous Biology
- Jason Lee, MD\*, Assistant Professor of Dermatology and Cutaneous Biology
- Kehua Li, MD\*, Adjunct Assistant Professor of Dermatology and Cutaneous Biology
- Joseph P. McFarland, MD, Assistant Professor of Dermatology and Cutaneous Biology
- Ellen Pfendner, PhD\*, Assistant Professor of Dermatology and Cutaneous Biology
- Franziska Ringpfeil, MD\*, Assistant Professor of Dermatology and Cutaneous Biology
- Fatima Rouan, PhD\*, Research Assistant Professor of Dermatology and Cutaneous Biology
- Takashi Tsuda\*, MD, Assistant Professor of Dermatology and Cutaneous Biology
- **Stephan S. Sollberg, MD,** Adjunct Research Assistant Professor of Dermatology and Cutaneous Biology<sup>14</sup>
- **Katsuto Tamai, MD, PhD,** Visiting Assistant Professor of Dermatology and Cutaneous Biology<sup>15</sup>

#### **INSTRUCTORS**:

Anne Marie C. Angeles, MD, Instructor of Dermatology and Cutaneous Biology
Gary L. Becker, MD, Instructor of Dermatology and Cutaneous Biology
Salvatore P. Calabro, MD, Instructor of Dermatology and Cutaneous Biology
David A. High, MD, Instructor of Dermatology and Cutaneous Biology
Olga Igoucheva, PhD\*, Instructor of Dermatology and Cutaneous Biology
Te-Cheng Pan, PhD\*, Instructor of Dermatology and Cutaneous Biology
Robin S. Scheiner, MD, Instructor of Dermatology and Cutaneous Biology
Toby Shaw, MD, Instructor of Dermatology and Cutaneous Biology
Barbara Vail, D.V.M., M.D.\*, Instructor of Dermatology and Cutaneous Biology
Kenneth Wasserman, MD, Instructor of Dermatology and Cutaneous Biology
Michele J. Ziskind, MD, Instructor of Dermatology and Cutaneous Biology

#### TJUH RESIDENTS IN DERMATOLOGY AND CUTANEOUS BIOLOGY

#### PGY-4

Judith Cenci, MD John Mulholland, MD Subhadra Sundaram, MD

#### PGY-3

Christine Ambro, MD Carmen Campanelli, MD Sarah Cash, MD John Wildemore, MD

#### PGY-2

Gina Ang, MD Reza Ghohestani, MD, PhD Aradna Saxena, MD Janelle Shield, MD

#### POST-DOCTORAL RESEARCH FELLOWS

Norihiro Fujimoto, MD, PhD
Hidetoshi Ito, MD
Qui-Jie Jiang, PhD
Dessislava Markova, PhD
Michael Naso, PhD
Marle Rocha DeQuadros, Ph.D.
Daniel Shurman, MD
Rosalyn Varki, MD
John Castorino, BS, Pre-doctoral Fellow
Hae Young Choi, MD, PhD, Visiting Scientist<sup>16</sup>

#### **ENDNOTES**:

- \* Denotes full-time faculty appointment at Jefferson Medical College
- President, Zagazig University, Cairo, Egypt
- 2 Director, Aomori Rosai Hospital, Hachenoke, Japan
- Professor Emeritus, Section of Dermatology, Institute of Occupational Health, University of Helsinki, Finland
- Professor, Department of Medical Biochemistry, University Hospital, Oulu, Finland
- Professor, Department of Dermatology, University of Florence, Italy
- 6 Primary appointment to the Department of Medicine, Jefferson Medical College
- Senior Lecturer and Head, Department of Dermatology, St. Jude's Medical Centre, Malta
- Professor, Department of Dermatology, Yonsei University School of Medicine, Seoul, Korea
- 9 Professor Emeritus, Dermatology at Oxford University, United Kingdom
- Professor and Chairman, Department of Obstetrics & Gynecology, University of Oulu, Finland
- Professor of Dermatology, University of Helsinki, Finland
- Research Director, Department of Dermatology, Hopital Saint-Louis, Paris, France
- Assistant Professor of Dermatology, Yonsei University Medical Center, Seoul, Korea
- 14 Chief, Department of Dermatology, Schwerin, Germany
- Associate Professor of Gene Therapy Sciences, University of Osaka, Japan
- Associate Professor, Department of Dermatology, Ewha Womans University Mokdong Hospital, Seoul, Korea

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program

#### PEER-REVIEWED PUBLICATIONS

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Pulkkinen, L., Choi, Y.W., Kljuic, A., **Uitto, J.,** and Mahoney, M.G.: Novel Member of the Mouse Desmoglein Gene Family: Dsg1-β: *Exp. Derm.* 12:11-19, 2003.

Matsuzaki, Y., Kon, A., Katsuto, T., Sawamura, D., **Uitto, J.,** and Hashimoto, I.: Keratinocyte Responsive Element 3 (KRE3): Analysis of a Keratinocyte-Specific Regulatory Sequence in the 230-kD Bullous Pemphigoid Antigen (BPAG1) Gene Promoter. *J. Invest. Dermatol.* 120:308-312, 2003.

Metz, B.J., Ruggeri, S.Y., Hsu, S., Reed, J.A., Ghohestani, A.S., **Uitto, J.,** and Ghohestani, R.F.: Linear IgA Dermatosis with IgA and IgG Autoantibodies to the 180 kDa Bullous Pemphigoid Antigen (BP180): Evidence for a Distinct Subtype. *Int. J. Derm.* (in press).

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

#### **SUBMITTED PUBLICATIONS:**

Nakano, H., Mauviel, A., Iozzo, R., and **Uitto, J.**: Interferon-γ Counteracts the Transcriptional Upregulation of Type VII Collagen Gene Expression Elicited by Transforming Growth Factor-β Evidence for Involvement of Stat-1 but not the Smad Pathway in Human Epidermal Keratinocytes.

Brittingham, R., Colombo, M., Ito, H., Steplewski, A., Birk, D.E., **Uitto, J.,** Fertala, A.J.: Single Amino Acid Substitutions in Procollagen VII Affect Early Stages of Assembly of Anchoring Fibrils.

Hata, D., Miyazaki, M., Seto, S., Furusho, K., Kadota, E., Nakano, A., Tamai, K., **Uitto, J.,** Moriyama, K., and Miyazaki, K.: Nephrotic Syndrome and Aberrant Expression of Laminin Isoforms in Glomerular Basement Membranes in an Infant with Herlitz Junctional Epidermolysis Bullosa.

Matsuzaki, Y., Jiang, Q.-J., Li, K., and **Uitto, J.**: Transcriptional Regulation of ABCC6 Gene Expression. Identification of a Novel Liver-Specific *Cis*-Element, Role of SP1, and Cytokine Modulation of the Promoter Activity.

Matsuzaki, Y., Nakano, A., Pulkkinen, L., and **Uitto, J.**: The ABCC6 Gene: Tissue-Specific Expression and Evidence for Alternative Splicing in the 3'-End of the mRNA.

Kaneko, T., Tamai, K., Matsuzaki, Y., Yamazaki, T., Nakano, H., Kon, A., Hashimoto, I., Hanada, K., Kaneda, Y., and **Uitto, J.**: Interferon-g Downregulates Expression of the 230-kD Bullous Pemphigoid Antigen Gene (BPAG1) via Novel Chimeric Sequences of ISRE and GAS in Epidermal Keratinocytes.

Brennan, D., Hu, Y., Corealle, D., Choi, Y.W., Joubeh, S., Kljuic, A., Wahl, J.K., Christiano, A.M., **Uitto, J.,** and Mahoney, M.: Differential Expression of Dsg1- $\beta$  and Dsg1- $\alpha$  in Mouse Stratified Epithelial Tissues.

Matsuzaki, Y., Jiang, Q-J., Li, K., and **Uitto, J.**: Transcriptional Regulation of ABCC6 Gene Expression. Identification of a Novel Liver-Specific *Cis*-Element, Role of Sp1, and Cytokine Modulation of the Promoter Activity.

Kalinin, A.E., Kalinin, A.E., Aho, M., <u>Uitto, J.</u>, and Aho, S.: Breaking the Connection: Caspase 6 Cleaves Periplakin Releasing the Intermediate Filament Binding Tail Region from the Actin Binding N-Terminal Domain.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

Pfendner, E., and **Uitto, J.:** Plectin Gene Mutations Cause Epidermolysis Bullosa with Pyloric Atresia

#### CHAPTERS IN BOOKS AND REVIEW ARTICLES

**Uitto, J.,** Richard, G.: Progress in Epidermolysis Bullosa: From Eponyms to Molecular Genetic Classification. <u>Clin. Dermatol.</u> (in press).

**Uitto, J.:** Clinical Implications of Basic Research on Heritable Skin Diseases. Abstracts, The IX International Congress of Dermatology, Beijing, China, May 19-22, 2004, Abstract 04.

Murrell, D.F., Turner, A., Moran, K., Pfendner, E., **Uitto, J.,** Lloyd, L., Klingberg, S.: Prenatal Diagnosis of Epidermolysis Bullosa in Australia. Abstracts of the Australasian College of Dermatologists 57<sup>th</sup> Annual Meeting, Sydney, New South Wales, Australia, May 16-19, 2004.

**Uitto, J.:** Epidermolysis Bullosa: The Expanding Mutation Database. (Commentary) J. Invest. Dermatol. (in press).

**Uitto, J.:** Structure & Function of the Skin. (CRS 203). The 62<sup>nd</sup> Annual Meeting of the American Academy of Dermatology, Washington, DC, February 7, 2004.

**Uitto, J.:** Pseudoxanthoma Elasticum – A Conective Tissue Disorder or a Metabolic Disease at the Genome/Environmental Interface? (Commentary). <u>J. Invest.</u> <u>Dermatol.</u> 122(3):ix-x, 2004.

Ringpfeil, F., and **Uitto, J.:** Pseudoxanthoma Elasticum. <u>Encyclopedic Reference of Genomics and Proteomics</u> (in press).

**Uitto, J.,** and Ringpfeil, F.: Heritable Disorders of Connective Tissue: The Paradigms of Ehlers-Danlos Syndrome and Pseudoxanthoma Elasticum. <u>In: Principles of Molecular Medicine</u> (Goldsmith, L., and Diaz, L.A., eds.) Second Ed., Humana Press, Totowa, N.J. (in press).

**Uitto, J.,** and Ringpfeil, F.: Ehlers-Danlos Syndrome – Molecular Genetics Beyond the Collagens. <u>J. Invest. Dermatol.</u> 122(4):XII, 2004.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

- **Uitto, J.,** Richard, G., and Christiano, A.M.: Molecular Genetics Of Epidermolysis Bullosa. <u>In:</u> Principles of Molecular Medicine. (Goldsmith, L., and Diaz, L.A., eds). Second Ed., Humana Press, Totowa, N.J. (in press).
- **Uitto, J.,** and Richard, G.: Progress in Epidermolysis Bullosa: Genetic Classification and Clinical Implications. In: Genetic Disorders of the Skin. <u>Am. J. Med. Genet.</u> Seminar. (in press).
- Richard, G., and **Uitto, J.:** Heritable Skin Disorders. <u>Encyclopedic Reference of Genomics and Proteomics.</u> (in press).
- **Uitto, J.,** Pfendner, E., and Jackson, L.C.: Probing the Fetal Genome: Progress Towards Non-Invasive Prenatal Diagnosis. *Trends Mol. Med.* 9:339-343, 2003.
- **Uitto, J.,** and Chu, M-L.: Elastic Fibers. <u>In:</u> Fitzpatrick's Dermatology in General Medicine (Freedberg, I.M., et al., eds), Sixth Edition, McGraw-Hill, New York, 2003, pp. 180-189.
- **Uitto, J.,** Pulkkinen, L., and Chu, M-L.: Collagen. <u>In:</u> Fitzpatrick's Dermatology in General Medicine (Freedberg, I.M., et al., eds), Sixth Edition, McGraw-Hill, New York, 2003, pp. 165-179.
- **Uitto, J.:** Collagen and Elastin & Their Alterations in Acquired and Inherited Skin Diseases: <u>In:</u> Structure & Function of the Skin (CRS 203). The 61<sup>st</sup> Annual Meeting of the American Academy of Dermatology, San Francisco, California, March 21 26, 2003.
- **Uitto, J.,** Ringpfeil, F., and Pulkkinen, L.: Heritable Disorders of Connective Tissue Ehlers-Danlos Syndrome, Pseudoxanthoma Elasticum and Cutis Laxa. <u>In: Dermatology</u> (Bologna, J.L., Jorizzo, J.L., and Rapini, R.P., eds.) Harcourt Publishers, London, 2003, pp. 1519-1530.

#### **ABSTRACTS**

Ghahestani, A.S., Nikbakht, N., Fry, L., Reunala, T., **Uitto, J.,** and Ghohestani, R.: Circulating IgA Autoantibodies to Epidermal Transglutaminase 3 Can Not Distinguish Dermatitis Herpetiformis from Celiac Disease. *J. Invest. Dermatol.* 122: Abstract 081, 2004.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

Nguyen, P., Bazzi, H., Kljuic, A., Vargas, J.H., Dmochowski, M., Barria, M., Ghohestani, R.F., Chan, L.S., **Uitto, J.,** Diaz, L.A., Christiano, A.M., Mahoney, M.G., and Nguyen, V.T.: Identification of a Novel Population of Pathogenic Autoantibodies Against Desmoglein 4 in Different Major Variants of Pemphigus. *J. Invest. Dermatol.* 122: Abstract 085, 2004.

Fujimoto, N., Terlizzi, J., Li, K., Aho, S., and **Uitto, J.:** Assembly of Elastic Fibers: Elastin and MFAP3 Interactions Determined by Yeast Two-Hybrid Genetic System: *J. Invest. Dermatol.* 122: Abstract 215, 2004.

Brennan, D.M., Hu, Y., Kljuic, A., **Uitto, J.,** Panteleyev, A., Christiano, A.M., Mahoney, M.G.: Complexity of Dsg1- $\alpha$ , - $\beta$ , and - $\gamma$  Expression in Diverse Mouse Tissues and During Hair Follicle Formation. *J. Invest. Dermatol.* 122: Abstract 222, 2004.

Terlizzi, J., Li, K., Aho, S., Fujimoto, N., Oyama, N., Hamada, T., Chan, I., McGrath, J.A., and **Uitto, J.:** Characterization of ECM-1 Protein Interactions by Yeast Two-Hybrid System. *J. Invest. Dermatol.* 122: Abstract 225, 2004.

Kalinin, A.E., Kalinin, A.E., Aho, M., **Uitto, J.,** and Aho, S.: Breaking the Connection: Caspase 6 Cleaves Periplakin Releasing the Intermediate Filament Binding Tail Region from the Actin Binding N-terminal Domain. *J. Invest. Dermatol.* 122: Abstract 437, 2004.

Richard, G., Ratajczak, P.A., Amin, S., Ilyas, H., Tesfaye Kedjela, A., Siegfried, E.C., and **Uitto, J.:** Netherton Syndrome: Novel and Recurrent Mutations in SPINK5 and Implications for Screening and Diagnosis. *J. Invest. Dermatol.* 122: Abstract 483, 2004.

Sprecher, E., Abu Sa'd, J., Pfendner, E., Indelman, M., Ciubutaro, D., Mizrachi, M., Lestringant, G., Pulkkinen, L., Richard, G., Kanaan, M., **Uitto, J.,** and Bergman, R.: Molecular Epidemiology of Epidermolysis Bullosa in Middle East Populations. *J. Invest. Dermatol.* 122: Abstract 493, 2004.

**Uitto, J.** and Pfendner, E.: Compound Heterozygosity for Unique In-Frame Insertion and Deletion Mutations in the Plectin Gene in a Mild Case of Epidermolysis Bullosa with Very Late Onset Muscular Dystrophy. *J. Invest. Dermatol.* 122: Abstract 513, 2004.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

- Pfendner, E. and **Uitto, J.:** Novel and Recurrent Mutations in the Keratin 5 and 14 Genes in Epidermolysis Bullosa Simplex: Implications for Genetic Counseling. *J. Invest. Dermatol.* 122: Abstract 516, 2004.
- Fratta, S., Ringpfeil, F., Terry, S., Terry, P., **Uitto, J.** and Pfendner, E.: dHPLC Screening Detects Novel and Recurrent Mutations in Pseudoxanthoma Elasticum. *J. Invest. Dermatol.* 122: Abstract 517, 2004.
- Jiang, Q., Matsuzaki, Y., Li, K. and **Uitto, J.:** Transcriptional Regulation of ABCC6 Gene Expression: Identification of Novel *Cis*-Elements, Role of Sp1, and Cytokine Modulation of the Promoter Activity. *J. Invest. Dermatol.* 122: Abstract 522, 2004.
- Colombo, M., Brittingham, R.J., Klement, J., Birk, D.E., **Uitto, J.,** and Fertala, A.: Procollagen VII Self-Assembly and its Perturbations in Dystrophic Epidermolysis bullosa. *J. Invest. Dermatol.* 122: Abstract 525, 2004.
- Ringpfeil, F., McGuigan, K., Fuchsel, L., Kozic, H., Lebwohl, M., and **Uitto, J.:** Autosomal Dominant Inheritance in Pseudoxanthoma Elasticum Revisited. *J. Invest. Dermatol.* 122: Abstract 551, 2004.
- Fuchsel, L., Kozic, H., McGuigan, K., Skvarka, C., Jacobson, M., **Uitto, J.** and Ringpfeil, F.: Genotype-Phenotype Correlation in 62 Patients with Pseudoxanthoma Elasticum. *J. Invest. Dermatol.* 122: Abstract 554, 2004.
- Ringpfeil, F., McGuigan, K., Kozic, H., and **Uitto, J.**: Pseudodominance in Pseudoxanthoma Elasticum A Common Phenomenon: Implications for Genetic Counseling. *Am. J. Human Genet.* 73: Abstract 710, 2003.
- **Uitto, J.,** Matsuzaki, Y., Terlizzi, J., Li, K., Leperi, D., Klement, J., and Pulkkinen, L.: Pseudoxanthoma Development of a Mouse Model by Targeted Ablation of ABCC6. *Am. J. Human Genet.* 73: Abstract 1649, 2003.
- Richard, G., Yi, L., Wasserman, D., Rouan, F., Jan, A.Y., Sybert, V.P., and **Uitto, J.**: Moleclar Studies in KID Syndrome Reveal a Mutational "Hot Spot" in Cx26, Genetic Heterogeneity and an Intriguing Phenotypic Variability. Abstracts, International Gap Junction Conference, St. John's College, University of Cambridge, Cambridge, United Kingdom, August 23 28, 2003, p. 119.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

- Rouan, F., Yi, L., **Uitto, J.,** and Richard, G.: Similar Mutations of Cx31 and Cx30.3 Impair Gap Junction Function, Dominantly Inhibit Wtcx31, and have Different Consequences for Cell Survival. Abstracts, International Gap Junction Conference, St. John's College, University of Cambridge, Cambridge, United Kingdom, August 23 28, 2003, p. 123.
- **Uitto, J.**: Molecular Biology of the Skin and the Environment: The Paradigm of Cutaneous Aging. Abstracts, First World Congress on Work-Related and Environmental Allergy, Helsinki, Finland, July 9-12, 2003, p. 51.
- Shu, D., Peritz, A.E., Youn, J., Gasparro, F.P., and **Uitto, J.**: 8-Methoxypsoralen Photoadducts Mapping of p53 Gene in the Fibroblasts Treated with PUVA. *J. Invest. Dermatol.* 121: Abstract 1132, 2003.
- Kljuic, A., Sundberg, J., Bazzi, H., Martinez-Mir, A., O'Shaugnessy, R., Djabali, K., Levy, M., Montagutelli, X., Ahmad, W., Aita, V.M., Gordon, D., Mahoney, M., Uitto, J., Whiting, D., Ott, J., Fisher, S., Gilliam, T., Morris, R., Panteleyev, A., and Christiano, A.M.: Desmoglein 4 is a Novel Desmosomal Cadherin with an Essential Role in Hair Follicle Keratinocyte Differentiation and is Mutated in the Lanceolate Mouse and Localized Hypotrichosis in Humans. *J. Invest. Dermatol.* 121: Abstract 0844, 2003.
- Markova, D., Zou, Y., Ringpfeil, F., Sasaki, T., Kostka, G., Timpl, R., **Uitto, J.,** and Chu, M.L.: Genetic Heterogeneity of Cutis Laxa: A Heterozygous Tandem Duplication within the Fibulin-5 Gene. *J. Invest. Dermatol.* 121: Abstract 0677, 2003.
- Matsuzaki, Y., Terlizzi, J., Li, K., Leperi, D., Klement, J., Pulkkinen, L., and **Uitto, J.**: Pseudoxanthoma Elasticum Development of a Mouse Model by Targeted Ablation of ABCC6. *J. Invest. Dermatol.* 121: Abstract 0672, 2003.
- Richard, G., Wasserman, D., Sundaram, S., Kozic, H., Itin, P., Lewanda, A., Antaya, R., Willoughby, C., and **Uitto, J.**: Genotype-Phenotype Correlations in KID Syndrome: Novel GJB2 Mutations and Association with Squamous Cell Carcinoma. *J. Invest. Dermatol.* 121: Abstract 0666, 2003.
- Pulkkinen, L., Meng, X., Klement, J., Leperi, D., Birk, D., Sasaki, T., Timpl, R., and **Uitto, J.**: Induced Apoptosis, and Altered Adhesion, Proliferation and Gene Expression of Keratinocytes in the Epidermis of Mice Deficient in the Laminin  $\gamma$ -2 Chain. *J. Invest. Dermatol.* 121: Abstract 0658, 2003.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

Kozic, H., Oldemeyer, B., Kusseluk, E., Saxena, A., **Uitto, J.,** and Ringpfeil, F.: Cardiovascular Disease in Pseudoxanthoma Elasticum is not Linked to Mutations in ABCC6. *J. Invest. Dermatol.* 121: Abstract 0642, 2003.

Rouan, F., Yi, L., **Uitto, J.,** and Richard, G.: pathogenic Mutations Affecting Cx31 and Cx30.3 Impair Gap Junction Function and Induce Cell Death. *J. Invest. Dermatol.* 121: Abstract, 0603, 2003.

Pfendner, E., Fratta, S., Sadowski, S., and **Uitto, J.**: dHPLC Based Mutation Detection for Dystrophic Epidermolysis Bullosa. *J. Invest. Dermatol.* 121: Abstract 0390, 2003.

Brennan, D., Hu, Y., Corealle, D., Choi, Y., Joubeh, S., Kljuic, A., Wahl, J., Zlotogorski, A., Christiano, A., **Uitto, J.,** Pulkkinen, L., and Mahoney, M.: Role of Novel Desmosomal Cadherin, Desmoglein 1-β, in Cell-Cell Adhesion and Blistering Disease. *J. Invest. Dermatol.* 121: Abstract 0251, 2003.

#### **AWARDS AND HONORS**

Invited Plenary Lecturer, IX International Congress of Dermatology, Beijing, China, May 19-22, 2004.

Honorary Professorship, Department of Dermatology, China Medical University, Shenyang, China, May 28, 2004.

Included in *Best Doctors in America*, 2003-2004.

President and Chair of the Organizing Committee, The 12<sup>th</sup> International Symposium on Basement Membranes, to be held at Jefferson Medical College, Philadelphia, Pennsylvania, June 16-19, 2005 (appointed in the 11<sup>th</sup> Symposium, Japan, March 6-9, 2003).

Plenary Speaker, First World Congress on Work-Related and Environmental Allergy, Finnish Institute of Occupational Health, Helsinki, Finland, July 9-12, 2003.

Honorary Member (Number 5), Scandinavian Connective Tissue Society, Appointed in the Third Swedish-Finnish Connective Tissue Meeting, Harjattula, Turku, Finland, September 26, 2003.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

#### VISITING PROFESSORSHIPS AND INVITED LECTURESHIPS

Visiting Professor, Department of Dermatology, Brown University School of Medicine, Providence, Rhode Island, January 7-8, 2004.

Invited Speaker, Course "Structure and Function of the Skin", 62<sup>nd</sup> Annual Meeting of the American Academy of Dermatology, Washington, D.C., February 7, 2004.

Visiting Scientist, University of Hawaii, Manoa, Oahu, Hawaii, February 12-13, 2004.

Visiting Lecturer, Division of Dermatology, Department of Medicine, University of California, San Diego, San Diego, California, February 26,2004.

Invited Discussant, Scientific Planning Retreat, National Institute of Arthritis and Musculoskeletal and Skin Diseases, National Institutes of Health, The Aspen Institute, Aspen Wye River, Maryland, March 28-30, 2004.

Seminar Speaker, Science Center, University of Pennsylvania, Philadelphia, Pennsylvania, April 15, 2004.

Visiting Scientist, The Smurfit Institute for Genetics, Trinity College, Dublin, Republic of Ireland, April 21 and 22, 2004.

Invited Speaker, The Annual Meeting of the Irish Association of Dermatologists, Armaugh, Northern Ireland, United Kingdom, April 23 and 24, 2004.

Invited Participant and Session Chair, Symposium on "Extracellular Matrix: Assembly, Functions and Role in Disease – A Tribute to Rupert Timpl". Max Planck Institute for Biochemistry, Martinsreid, Germany, April 25-27, 2004.

Invited Speaker, Department of Dermatology, Peking University First Hospital Skin and STD Center, Peking University, Beijing, China, May 20, 2004.

Guest Lecturer, Department of Dermatology, Fourth Military Medical University, National Key Discipline of Dermatology and Venereology Center of People's Liberation Army of China, Xijing Hospital, Xi'an, China, May 25, 2004.

Invited Speaker, Department of Dermatology, No. 1 Hospital of the China Medical University, Shenyang, China, May 27, 2004.

Professor and Chair, Department of Dermatology and Cutaneous Biology Professor, Department of Biochemistry and Molecular Pharmacology Director, Jefferson Institute of Molecular Medicine Program Director, Dermatology Residency Training Program (continued)

Invited Speaker, The 4<sup>th</sup> Resident Retreat for Future Physician-Scientists, Airlie Center, Warrenton, Virginia, June 4-6, 2004.

Grand Round Speaker, Department of Dermatology, Johns Hopkins University School of Medicine, Baltimore, Maryland, June 9, 2004.

Invited Speaker, Third European Symposium, Elastin-2004, University of Manchester, Manchester, United Kingdom, June 30 – July 3, 2004.

Invited Speaker, University of Hawaii, Manoa, Oahu, Hawaii, January 26-28, Invited Speaker and Session Chair, Gordon Conference on "Epithelial Differentiation and Keratinization", Tilton, New Hampshire, July 13-18, 2003.

Consultant and Invited Lecturer, Avon Products, Inc., Suffern, New York, September 5, 2003.

Invited Speaker, Frontiers of Science Seminars, Biocity, University of Turku, Finland, September 25, 2003.

Keynote Lecturer, The Third Swedish-Finnish Connective Tissue Meeting, Harjattula, Turku, Finland, September 26 & 27, 2003.

Guest Lecturer, Campus-Wide Graduate Seminars, Drexel University, Philadelphia, Pennsylvania, October 3, 2003.

Keynote Speaker, Second International Symposium on Bullous Diseases: What is New? Department of Dermatology, Heinrich-Heine University, Düsseldorf, Germany, October 11, 2003.

Invited Speaker, NIH Sponsored: Genetic Diseases Impacting Processes of Aging and Longevity. The 56<sup>th</sup> Annual Scientific Meeting of the Gerantological Society of America, San Diego, California, November 21-23, 2003.

#### SIRPA AHO, PH.D.

Research Associate Professor of Dermatology and Cutaneous Biology, Department of Dermatology and Cutaneous Biology Research Associate Professor of Pathology, Anatomy and Cell Biology (secondary appointment)

#### **PUBLICATIONS:**

Kazerounian, S. and **Aho, S.** Characterization of periphilin, a widespread, highly insoluble nuclear protein and potential constituent of the keratinocyte cornified envelope. J. Biol. Chem. 278: 36707-36717, 2003.

**Aho, S.** and Kazerounian, S. Molecular interactions of periplakin in epithelial cells. Recent Res. Devel. Cell. Biochem., 1: 131-141, 2003. Transworld Research Network, Kerala, India.

**Aho, S.** Many faces of periplakin: domain-specific antibodies detect the protein throughout the epidermis explaining its multiple protein-protein interactions. Cell Tissue Res., 316: 87-97, 2004.

**Aho, S.** Soluble form of Jagged1: Unique product of epithelial keratinocytes and a regulator of keratinocyte differentiation. J. Cell. Biochem., 92:1271-1281, 2004.

**Aho, S.** Plakin proteins are coordinately cleaved during apoptosis but preferentially through the action of different caspases. Exp. Dermatol. In press.

**Aho, S.,** Li, K., Ryoo, Y., McGee, C., Ishida-Yamamoto, A., Uitto, J. and Klement, JF. Periplakin gene targeting reveals a constituent of the cornified cell envelope dispensable for normal mouse development. Mol. Cell. Biol. 24:6410-6418, 2004.

Kalinin, AE, Kalinin, AE, Aho, M., Uitto, J. and **Aho, S.** Breaking the connection: Caspase 6 disconnects intermediate filament binding domain of periplakin from its actin binding N-terminal region. J. Invest. Dermatol. In press.

#### **GRANT SUPPORT:**

"Hemidesmosomal Protein Linkage Map by Two-hybrid System". NIH/NIAMS: 1RO1-AR44833, \$121,691/y.

Principal investigator: Sirpa Aho, Ph.D.

#### **PARTICIPATION IN SCIENTIFIC MEETINGS:**

Gordon Research Conference, Epithelial Differentiation and Keratinization. Tilton School, Tilton, NH, July 13-18, 2003.

The 65<sup>th</sup> Annual Meeting of the Society for Investigative Dermatology, April 28-May 1, 2004, Providence, RI.

#### SIRPA AHO, PH.D.

Research Associate Professor of Dermatology and Cutaneous Biology, Department of Dermatology and Cutaneous Biology Research Associate Professor of Pathology, Anatomy and Cell Biology (secondary appointment) (continued)

#### **CONGRESS ABSTRACTS:**

**Aho, S.** Novel components involved in the differentiation of epidermal keratinocytes. Gordon Research Conference, Epithelial Differentiation and Keratinization. Tilton School, Tilton, NH, July 13-18, 2003.

Fujimoto, N., Terlizzi, J., Li, K., **Aho, S.** and Uitto, J. Assembly of elastic fibers: elastin and MFAP3 interactions determined by yeast two-hybrid genetic system. J. Invest. Dermatol. 122,A36, 2004. Abstract #215.

Terlizzi, J., Li, K., **Aho, S.,** Fujimoto, N., Oyama, N., Hamada, T., Chan, I., McGrath, JA. and Uitto, J. Characterization of ECM-1 protein interactions by yeast two-hybrid system. J. Invest. Dermatol. 122,A38, 2004. Abstract #225.

**Aho, S.** Plakin proteins are coordinately cleaved during apoptosis but preferentially through the action of different caspases. J. Invest. Dermatol. 122,A73, 2004. Abstract #436.

Kalinin, AE., Kalinin, AE., Aho, M., Uitto, J. and **Aho, S.** Breaking the connection: Caspase 6 cleaves periplakin releasing the intermediate filament binding tail region from the actin binding N-terminal domain. J. Invest. Dermatol. 122,A73, 2004. Abstract #437.

#### GINA ANG, MD

Resident (PGY-3), Department of Dermatology and Cutaneous Biology

#### **PUBLICATIONS:**

Ang G, Lee JB. "A Pigmented Verrucous Plaque on the Cheek." *Arch Dermatol*. Accepted for publication.

Ang G. "History of Skin Transplantation." Clinics in Dermatology. Accepted for publication.

Ang G, Werth V. "Combination Antimalarials in the Treatment of Cutaneous Dermatomyositis: A Restrospective Study." Submitted.

#### **PRESENTATIONS:**

Ang G. "Dermatitis Herpetiformis." Philadelphia Dermatological Society, Philadelphia, 10/17/03.

Ang G, Hyde PM, Lee JB. "Congenital Linear Atrophoderma." International Society of Dermatopathology. Washington D.C., 2/4/04

Ang G, Werth V. "Combination Antimalarials in the Treatment of Cutaneous Dermatomyositis: A Restrospective Study." Philadelphia Dermatological Society: Johnson-Beerman Award, Philadelphia. 4/7/04.

#### **AWARDS:**

Johnson-Beerman Award, for clinical research. 4/7/04. Ang G, Werth V. "Combination Antimalarials in the Treatment of Cutaneous Dermatomyositis: A Restrospective Study."

#### MON-LI CHU, PH.D.

Professor and Vice-Chair of Research, Department of Dermatology and Cutaneous Biology

#### **PUBLICATIONS**:

Pan, T.-C., Zhang, R.-Z., Sudano, D.G., Marie, S.K., Bönnemann, C.G. and Chu, M.-L. (2003) New molecular mechanism for Ullrich congenital muscular dystrophy: A heterozygous in-frame deletion in the COL6A1 gene causes a severe phenotype. Am. J. Hum. Genet. 73:355-369.

Tsuda, T., Gao, E., Evangelisti, L., Markova. D., Ma, X.L., and Chu, M.-L. (2003) Post-ischemic myocardial fibrosis occurs independent of hemodynamic changes. Cardiovas. Res. 59:926-993.

Tsuda, T., Markova, D., Wang, H., Evangelisti, L., Pan, T.-C., and Chu, M.-L. (2004) The zinc finger protein Zac1 is expressed in chondrogenic sites of the mouse. Dev. Dyn. 229:340-348.

Chu, M-L., and Tsuda, T. (2004) Fibulins in development and heritable disease. Birth Defects Res Part C Embryo Today. 72:25-36.

Kassner, A., Tiedemann, K., Notbohm, H., Ludwig, T., Morgelin, M., Reinhardt, D.P., Chu, M.-L., Bruckner, P., Grässel, S. (2004) Molecular structure and interaction of recombinant human type XVI collagen. J Mol Biol 339:835-853.

Lampe, A.K., Dunn, D.M., von Niederhausern, A.C., Hamil, C., Aoyagi, A., Laval, S.H., Chu, M.-L., Swoboda, K., Muntoni, F., Bönnemann, C.G., Flanigan, K.M., Bushby, K.M.D.; Weiss, R.B. Automated genomic sequence analysis of the three collagen VI genes: Applications to Ullrich congenital muscular dystrophy and Bethlem myopathy. Submitted.

#### PARTICIPATION IN SCIENTIFIC MEETINGS:

43<sup>rd</sup> Annual Meeting of the American Society for Cell Biology. December 13-17, 2003, San Francisco, CA.

Extracellular matrix: assembly, functions and role in disease, April 25-27, 2004. Max Planck Institute of Biochemistry, Martinsried near Munich, Germany.

#### **DEPARTMENTAL COMMITTEES**

Coordinator of "Science Lecture Series" and "Research in Progress"

#### MON-LI CHU, PH.D.

Professor and Vice-Chair of Research, Department of Dermatology and Cutaneous Biology (continued)

#### **PROFESSIONAL ORGANIZATIONS**

American Society for Biochemistry and Molecular Biology American Association for the Advancement of Science International Society for Matrix Biology American Society for Matrix Biology Alexander von Humboldt Association of America American Society for Cell Biology

#### MARLENE ROCHA DE QUADROS, PH. D.

Post-Doctoral Fellow, Department of Dermatology and Cutaneous Biology

#### **PUBLICATIONS:**

Quadros, M.R.D., Peruzzi, F., Kari, C., Rodeck, U. "Complex regulation of activation in normal and malignant keratinocytes. *Cancer Research*, 64:3934-3939, 2004.

Belloni, G., Ferrero, D., Carbone, A., Quadros M.R., Gramigni, C., Prati, A., Davidson, R., Mioli, P., Dughera, L., Emmanueli, G., Rodeck, U. "Inhibition of cell survival and invasive potential of colorectal carcinoma cells by tyrosine kinase inhibitor STI571. Cancer Biology and Therapy (In Press).

Landers, M.C., Dugger, N., Quadros, M., Hoffman, P.M., Gaulton, G.N. "Neuropathogenic murine leukemia virus TR1.3 induces selective syncytia formation of brain capillar endothelium. **Virology**, **321** (1): **57-64**, **2004**.

#### **POSTER PRESENTATIONS:**

Quadros, M.R., Peruzzi, F., Kari C., Rodeck, U. "Complex regulation of STAT3 activation in normal and malignant keratinocytes". Molecular Targets and Cancer Therapeutics, Boston, Massachussets, November 17-21, 2003.

Ren Q., Kari C., Quadros, M.R., Dicker A. "Extracellular matrix composition modulates radiation sensitivity induced by EGFR blockade in transformed keratinocytes". Molecular Targets and Cancer Therapeutics, Boston, Massachussets, November 17-21, 2003.

#### NORIHIRO FUJIMOTO, MD, PHD

Post-Doctoral Research Fellow, Department of Dermatology and Cutaneous Biology

#### **PUBLICATIONS:**

**Fujimoto N**, Tajima S. Advanced glycation end product (AGE)-immunoreactive materials in chronic prurigo patients receiving a long-standing haemodialysis. Br J Dermatol. 2004, 150; 757-60.

**Fujimoto N**, Akagi A, Tajima S. Expression of 67-kDa elastin receptor in annular elastolytic giant cell granuloma: elastin peptides induce monocyte-derived dendritic cells or macrophages to form granuloma in vitro. Exp Dermatol. 2004, 13; 179-84.

Mieno H, **Fujimoto N**, Tajima S. Eruptive vellus hair cyst in patients with chronic renal failure. Dermatology. 2004, 208; 67-9.

Miura Y, **Fujimoto N**, Komatsu T, Tajima S, Kawada A, Saito T, Fujii N. Immunohistochemical study of chronological and photo-induced aging skins using the antibody raised against D-aspartyl residue-containing peptide. J Cutan Pathol. 2004, 31; 51-6.

**Fujimoto N**, Tajima S. Extensive fixed drug eruption due to the Japanese herbal drug "kakkonto". Br J Dermatol. 2003, 149; 1303-5.

**N Fujimoto**, J Terlizzi, K Li, S Aho, and J Uitto. Assembly of elastic fibers: elastin and MFAP3 interactions determined by yeast two-hybrid genetic system. J Invest Dermatol. 2004, 122; A36

J Terlizzi, K Li, S Aho, **N Fujimoto**, N Oyama, T Hamada, I Chan, JA Mc Grath and J Uitto Characterization of ECM-1 protein interactions by yeast two-hybrid system. J Invest Dermatol. 2004, 122; A38

#### PARTICIPATION IN SICENTIFIC MEETINGS AND PRESENTATIONS:

Assembly of elastic fibers: elastin and MFAP3 interactions determined by yeast two-hybrid genetic system

**N Fujimoto**, J Terlizzi, K Li, S Aho, and J Uitto The 65th Annual Meeting of the SID, Providence, April 28- May 1, 2004

Characterization of ECM-1 protein interactions by yeast two-hybrid system J Terlizzi, K Li, S Aho, **N Fujimoto**, N Oyama, T Hamada, I Chan, JA Mc Grath and J Uitto The 65th Annual Meeting of the SID, Providence, April 28- May 1, 2004

#### **PROFESSIONAL ORGANIZATIONS**

The Japanese Society for Investigative Dermatology Japanese Dermatological Association

#### REZA F. GHOHESTANI, MD, PH.D.

Research Assistant Professor, Department of Dermatology and Cutaneous Biology

#### **PUBLICATIONS:**

Metz BJ, Ruggeri SY, Hsu S, Reed JA, Ghohestani AS, Uitto J, **Ghohestani RF**. Linear IgA dermatosis with IgA and IgG autoantibodies to the 180 kDa bullous pemphigoid antigen (BP180): evidence for a distinct subtype. Int J Dermatol. 2004;43(6):443-6.

Hisamatsu Y, Nishiyama T, Amano S, Matsui C, **Ghohestani R**, Hashimoto T. Usefulness of immunoblotting using purified laminin 5 in the diagnosis of anti-laminin 5 cicatricial pemphigoid. J Dermatol Sci. 2003;33(2):113-9.

**Ghohestani RF**, Rotunda SL, Hudson B, Gaughan WJ, Farber JL, Webster G, Uitto J. Crescentic glomerulonephritis and subepidermal blisters with autoantibodies to alpha5 and alpha6 chains of type IV collagen. Lab Invest. 2003;83(5):605-11.

#### **PRESENTATIONS:**

Invited speaker: RF Ghohestani. Role of laminin-5 in cell-matrix adhesion. University of Southern California, July 2003

Invited speaker: RF Ghohestani. What's new in immunodermatology. The International Geographic Congress, Shiraz, Nov. 2003.

Invited speaker: RF Ghohestani. Development of a novel mouse model for pemphigus vulgaris by DNA immunization. The College of Physicians of Philadelphia, Philadelphia, April 2004.

Invited faculty: RF Ghohestani. New autoimmune skin syndromes. 62rd Annual Meeting of the American Academy of Dermatology, Washington DC, Feb. 2004.

Oral presentation: RF Ghohestani, M. Barria, AS Ghahestani, J Uitto. Development of a mouse model for pemphigus vulgaris, a life threatening skin disease. 62rd Annual Meeting of the American Academy of Dermatology, Washington DC, Feb. 2004.

Poster presentation: RF Ghohestani, M. Barria, G. Webster, A. Claudy, J. Uitto. The a5 and a6 chains of type IV collagen are the target of IgG and IgA autoantibodies in a novel autoimmune disease characterized by crescentic glomerulonephritis and sub-epidermal blisters. 62rd Annual Meeting of the American Academy of Dermatology, Washington DC, Feb. 2004.

Oral presentation: AS Ghahestani, N Nikbacht, L Fry, T Reunala, J Uitto, RF Ghohestani. Circulating IgA autoantibodies to epidermal transglutaminase 3 can not distinguish dermatitis herpetiformis from celiac disease. The Society for Investigative Dermatology, Rhode Island, May 2004.

#### REZA F. GHOHESTANI, MD, PH.D.

Research Assistant Professor, Department of Dermatology and Cutaneous Biology (continued)

Oral presentation: P Nguyen, H Bazzi, A Klujic, JH Vargas, M Dmochowski, M Barria, RF Ghohestani, LS Chan, J Uitto, LA Diaz, AM Christiano, MG Mahoney and VT Nguyen. Identification of novel population of pathogenic autoantibodies against desmoglein 4 in different major variants of pemphigus. The Society for Investigative Dermatology, Rhode Island, May 2004.

#### **HONORS/SPECIAL RECOGNITIONS:**

Stelwagon Award in Basic Sciences, The College of Physicians of Philadelphia, April 2004

Junior Faculty Award. The American Association of Immunologists, 2003

Editor, European Journal of Dermatology

Member, NIDCR(NIH) Review Panel

#### **MEMBERSHIP IN PROFESSIONAL ORGANIZATIONS:**

American Association of Immunologists, FASEB, French Society of Dermatology, American College of Surgeons, American Medical Association, Thomas Bond Society (Pennsylvania Hospital), PA Medical Society, The Society for Investigative Dermatology, European Society for Dermatological Research.

#### DAVID J. HERRICK, PH.D.

Research Associate Professor, Department of Dermatology and Cutaneous Biology

#### **PUBLICATIONS:**

Louneva, N., Saitta, B., Herrick, D. J., Jimenez, S. A.: Transcriptional inhibition of type I collagen gene expression in scleroderma fibroblasts by the antineoplastic drug ecteinascidin 743. J. Biol. Chem. <u>278</u>: 40400-40407, 2003.

Liao, B., Meera, P., Charles, S., Hu, Y., Herrick, D. J., Brewer, G. Targeted knockdown of the RNA-binding protein CRD-BP promotes cell proliferation via an IGF-II-dependent pathway. Submitted for publication.

Herrick, D.J., Wang, D., Uitto, J.: Identification and functional analysis of RNA-binding proteins which interact with the 3'-UTRs of the  $\alpha_1(I)$  and  $\alpha_1(III)$  procollagen mRNAs. (Presented at the Eukaryotic mRNA Processing meeting, Cold Spring Harbor Laboratories, 2003.)

Castorino, J.J., Brewer, G.A., Herrick, D.J: c-myc coding region determinant binding proteins: effects on mRNA localization, stability, and translation. (Presented at the Eukaryotic mRNA Processing meeting, Cold Spring Harbor Laboratories, 2003.)

Herrick, D.J., Wang, D., Uitto, J.: Functional analysis of the  $\alpha_1(I)$  procollagen 5'-stem loop and 3'-UTR: Role of the  $\alpha$ CP, AUF1 and PABP RNA-binding proteins. (To be presented at the Translational Control Meeting, Cold Spring Harbor Laboratories, 2004.)

Castorino, J.J., Brewer, G.A., Herrick, D.J: Functional dissection of the c-myc coding region determinant. (To be presented at the Translational Control Meeting, Cold Spring Harbor Laboratories, 2004.)

#### TATYANA R. HUMPHREYS, M.D.

Clinical Associate Professor, Department of Dermatology and Cutaneous Biology Chief of Cutaneous Surgery, Jefferson Dermatology Associates

#### **RESEARCH PUBLICATIONS, PEER-REVIEWED:**

Covello S, **Humphreys T**, Lee J. A case of extraskeletal osteosarcoma with metastasis to the skin. J Am Acad Dermatol 2003;49:124-7.

Linder J and **Humphreys TR**. Surgical pearl: Immobilization of the helical rim for postauricular procedures. J Am Acad Dermatol 2004; 50:98.

**Humphreys TR**, Finkelstein D, Lee JB. Superficial leiomyosarcoma treated with Mohs micrographic surgery. Dermatol Surg 2004; 30 (1):108-112.

Wildemore J, Lee JB, **Humphreys TR.** Mohs Micrographic Surgery and Malignant Eccrine Neoplasms . Accepted for publication, Dermatologic Surgery 2004.

#### **EDITORIALS, REVIEWS, AND BOOK CHAPTERS:**

**Humphreys TR**. Laser hair removal in pigmented skin. Skin Med 2004; in press.

**Humphreys TR**. <u>Skin Cancer</u>. In Conn's Current Therapy 2004. WB Saunders Co, Elsevier Inc, Philadelphia PA. pp827-30.

#### **Alternative Media**

**Humphreys TR**. Ear lobe problems. In Women Doctors Guide to Health and Healing. Oxmoor House 2003, Birmingham, AL. pp 116, 499.

**Humphreys TR**." 'Face Creams'. Action News, Channel 6, 2/25/04.

**Humphreys TR.** "Better than Botox?", Action News, Channel 6, 5/5/04

Humphreys TR. "Skin cancer recognition", Channel 10 Live, 5/18/04

**Humphreys TR**. Skin Cancer screening, Channel 10 News, 5/19/04

**Humphreys TR**. Sunfree tanning: "Orange you glad you're tan". US News and World Report, 6/21/04: page D11

#### TATYANA R. HUMPHREYS, M.D.

Clinical Associate Professor, Department of Dermatology and Cutaneous Biology Chief of Cutaneous Surgery, Jefferson Dermatology Associates (continued)

#### **Lectures by Invitation**

October 2003 ." Mohs micrographic surgery and malignant eccrine tumors". American College of Mohs Micrographic Surgery and Cutaneous Oncology Meeting, New Orleans.

October 2003 " Challenges in Reconstruction: masters panel". American College of Mohs Micrographic Surgery and Cutaneous Oncology Meeting, New Orleans.

October 2003 "Mohs micrographic surgery: unusual applications". Philadelphia Dermatologic Society Meeting, Philadelphia.

April 2004." Skin Cancer". Foundations of Clinical Medicine Course. Jefferson Medical College, Philadelphia.

June 2004 "Restylane and other new facial fillers", Pennsylvania Academy of Otorhinolaryngology, Head and Neck Surgery, Philadelphia PA.

#### **Organizing Roles in Scientific Meetings**

June 2005-Current Co-Chair, Laser symposium of the World Congress of Aesthetic Dermatology, Paris, France

#### MEMBERSHIP IN PROFESSIONAL AND SCENTIFIC SOCIETIES:

#### National Societies:

Fellow, American Academy of Dermatology; 1994 to present

Fellow, American Society of Dermatologic Surgery; 1995-present

Fellow, American College of Mohs Micrographic Surgery and Cutaneous

Oncology; 1995-present

Member, Society for Investigative Dermatology; 1995 to present

#### Local Societies:

Philadelphia Dermatologic Society, 1997-present Philadelphia Facial Plastics Society, 1998-present

#### **Editorial Positions**

2000 to present Editorial board, Cosmetic Dermatology 2003 to present Contributing editor, Dermatologic Surgery

2004 to present Editorial board, Journal of the American Academy of

Dermatology

2004 to present Editorial board, Skin Med

Guest editor,' Lasers in Dermatology' in Clinics in Dermatology

## **OLGA IGOUCHEVA, PH.D.**

Instructor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Pierce E.A., Liu Q., <u>Igoucheva O.</u>, Omarrudin R., Ma H.C., Diamond S. and Yoon K. Oligonucleotide-directed single base DNA alterations in mouse embryonic stem cells. *Gene Therapy*, 10, 24-33, 2003.

<u>Igoucheva, O.</u>, Alexeev, V., Pryce, M. and Yoon, K. Transcription affects formation and processing of intermediates in oligonucleotide-mediated gene alteration. *Nucleic Acids Research*, 31, 2659-2670, 2003.

Yoon, K., Alexeev, V. and <u>Igoucheva</u>, <u>O.</u> Biased gene repair needs unbiased review. *Correspondence in Nature Review Genetics*, 4, 279-689, 2003.

<u>Igoucheva, O.</u> and Yoon, K. Gene targeting by oligonucleotides in keratinocytes. *Epidermal Cell Protocols*, Methods and Protocols, ed. Turksen, K. Humana Press, 289, 287-302, 2004.

<u>Igoucheva. O.</u>, Alexeev, V. and Yoon, K. Site-directed mutagenesis and targeted gene correction. *Current Molecular Medicine*, 4(5), 445-463, 2004.

<u>Igoucheva</u>, O., Alexeev, V. and Yoon, K. Mechanism of gene repair open for discussion. *Gene Therapy* (submitted) 2004.

<u>Igoucheva, O.</u>, Alexeev, V. and Yoon, K. Cellular response to exogenous DNA and its implication in gene targeting. *Proc. Natl. Acad. Sci. USA* (submitted) 2004.

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

February, 2004 Pachyonychia Congenita Project Retreat, Park City, UT, USA <u>Invited speaker</u>: "Oligonucleotide-mediated gene repair: Potential applications to human skin disorders".

## QIUJIE JIANG, PhD

Post-Doctoral Research Fellow, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Q Jiang, Y Matsuzaki, K Li and J Uitto. Transcriptional regulation of ABCC6 gene expression: identification of novel cis-elements, role of Sp1, and cytokine modulation of the promoter activity. *The Journal of Investigative Dermatology*, vol 122(3): A87, 2004

Transcriptional regulation of ABCC6 gene expression: identification of novel cis-elements, role of Sp1, and cytokine modulation of the promoter activity. Q Jiang, Y Matsuzaki, K Li and J Uitto. *The 65<sup>th</sup> Annual Meeting of the Society for Investigative Dermatology*. Providence, Rhode Island. April 28-May 1, 2004

## YOUNG C. KAUH, M.D.

Professor and Senior Vice-Chairman, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Cash, S. & Kauh, Y.: Zosteriform sarcoidosis.. Submitted for publication. J Am Acad Dermatol.

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

"Zoteriform sarcoidosis" at Gross & Microscopic Dermatology Symposium in 62<sup>nd</sup> Annual Convention of American Dermatological Meeting, February 7, 2004

"Rhus contact dermatitis and its management" at Annual Scientific Meeting of Yonsei University Medical School Alumni Association, Washington DC, August 6, 2003

"En coup de sabre" at Philadelphia Dermatological Society Meeting, October 17, 2003

"Hansen's disease" at Philadelphia Dermatological Society Meeting, October 17, 2003

"Eccrine hydrocystoma" at Philadelphia Dermatological Society Meeting, October 17, 2003

"Parapsoriasis lichenoides" at Philadelphia Dermatological Society Meeting, October 17, 2003

"Zosteriform sarcoidosis" at Philadelphia Dermatological Society Meeting, October 17, 2003

## **HONORS/SPECIAL RECOGNITIONS:**

Second Young C Kauh Clinical Dermatologic Lectureship by Dr. Samuel Moschella, Professor of Dermatology, Harvard Medical School, November 19, 2003

#### **DEPARTMENTAL COMMITTEES:**

Faculty Appointment and Promotions committee

## **UNIVERSITY COMMITTEES**:

Member of Professorial Global Advisory Board

## **PROFESSIONAL ORGANIZATIONS**:

Fellow of American Academy Dermatology Member of Philadelphia Dermatological Society

#### JOHN F. KLEMENT, PHD

Research Assistant Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Naso M, Uitto J, Klement JF: Transcriptional control of the mouse Col7a1 gene in keratinocytes: basal and transforming growth factor-beta regulated expression. J Invest Dermatol 2003, 121:1469-1478

Aho S, Li K, Ryoo Y, McGee C, Ishida-Yamamoto A, Uitto J, Klement JF: Periplakin gene targeting reveals a constituent of the cornified cell envelope dispensable for normal mouse development. Mol Cell Biol 2004, 24:6410-6418

Colombo M, Brittingham RJ, Klement JF, Majsterek I, Birk DE, Uitto J, Fertala A: Procollagen VII self-assembly depends on site-specific interactions and is promoted by cleavage of the NC2 domain with procollagen C-proteinase. Biochemistry 2003, 42:11434-11442

Meng X, Klement JF, Leperi DA, Birk DE, Sasaki T, Timpl R, Uitto J, Pulkkinen L: Targeted inactivation of murine laminin gamma2-chain gene recapitulates human junctional epidermolysis bullosa. J Invest Dermatol 2003, 121:720-731

## **UNIVERSITY COMMITTEES:**

Institutional Animal Care and Use Committee

## **PROFESSIONAL ORGANIZATIONS:**

American Association for Microbiology (ASM)
American Association for the Advancement of Science (AAAS)

#### JASON B. LEE, MD

Clinical Assistant Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Ambro C, Lee JB. Multiple basal-cell carcinomas associated with a port-wine stain. Submitted to Cutis 8/04

Divers AK, Correale D, Lee JB. Keratoacanthoma centrifugum marginatum: a diagnostic and therapeutic challenge. Cutis 2004; Vol 73: 257-62.

Cenci JA, Covello SP, Lee JB. A painless slowly enlarging tumor of the leg. *J Am Acad Dermatol* 2004; Vol 50: 159-61.

Humphreys TR, Finkelstein DH, Lee JB. Superficial leiomyosarcoma treated with Mohs micrographic Surgery. *Dermatologic Surgery* 2004; vol 30: 108-112.

Hwang LY, Richard G, Lee JB, Richard G, Uitto J, Hsu S. Type 1 segmental manifestation of Hailey-Hailey disease. *J Am Acad Dermatol* 2003; Vol 49: 712-714.

Santoro AF, Rezac MA, Lee JB. Current trend in ivermectin usage for scabies. *J Drugs Dermatol* 2003; Vol 2:397-401.

Covello SP, Humphreys T, **Lee JB**. A case of extraskeletal osteosarcoma metastasis to the skin. *J Am Acad Dermatol* 2003; Vol 49: 124-127.

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETING AND PRESENTATIONS:

International Society of Dermatopathology Meeting in Washington D.C. Free Communications. Congenital Linear Atrophoderma. Authors: Ang G, Lee JB. 2/04

The annual meeting of the American Academy of Dermatology 2004 in Washington D.C. Poster Session: Multiple basal-cell carcinomas associated with a port-wine stain. Authors: Ambro C, **Lee JB**.

## **UNIVERSITY COMMITTEES:**

JUP Clinical care committee JUP Risk management committee

## JASON B. LEE, MD

Clinical Assistant Professor, Department of Dermatology and Cutaneous Biology (continued)

## **PROFESSIONAL ORGANIZATIONS:**

Alpha Omega Alpha (A $\Omega$ A) Honor Medical Society

American Academy of Dermatology

American Society of Dermatopathology

International Society of Dermatopathology

Philadelphia Dermatological Society (11/02)

Pennsylvania Academy of Dermatology (1/03)

Pennsylvania Regional Dermatopathology Society (10/02)

Korean Dermatology Society (3/03)

Dermatology Foundation (7/03)

American Academy of Dermatology: Computer and Informatics Task Force (2/04)

## KEHUA LI, MD

Assistant Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Kaithamana S, Fan JL, Memar K, Li K, Uitto J, Seetharamaiah S, Prabhakar BS. Relevance of differential immunogenicity of human and mouse recombinant desmoglein-3 for the induction of acantholytic autoantibodies in mice. *Clin. Exp. Immunol.* 132:16-23, 2003

Aho S, Li K, Ryoo Y, McGee C, Ishida-Yamamoto A, Uitto J, Klement J. Periplakin gene targeting reveals a constituent of the cornified cell envelop dispensable for normal mouse development. *Molecular and Cellular Biology*, July 2004

Fujimoto N, Terlizzi J, **Li K**, Aho S, Uitto J. Assembly of elastic fibers: elastin and MFAP3 interactions determined by yeast two-hybrid genetic system. The 65<sup>th</sup> Annual Meeting of the Society for Investigative Dermatology. April 28 – May 1, 2004. *J. Invest. Dermatol.* 122:A36, 2004.

Terlizzi J, **Li K**, Aho S, Fujimoto N, Oyama N, Chan I, MaGrath JA, Uitto J. Characterization of ECM-1 protein interactions by yeast two-hybrid system. The 65<sup>th</sup> Annual Meeting of the Society for Investigative Dermatology. April 28 – May 1, 2004. *J. Invest. Dermatol.* 122:A38, 2004.

Jiang Q, Matsuzaki Y, **Li K**, Uitto J. Transcriptional regulation of ABCC6 gene expression: identification of novel *cis*-element, role of Sp1, and cytokine modulation of the promoter activity. The 65<sup>th</sup> Annual Meeting of the Society for Investigative Dermatology. April 28 – May 1, 2004. *J. Invest. Dermatol.* 122:A87, 2004

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

The  $65^{th}$  Annual Meeting of the Society for Investigative Dermatology. Washington, DC, April 28 - May 1, 2004

The IX International Congress of Dermatology, Beijing, China, May 18 -22, 2004.

## **HONORS/SPECIAL RECOGNITIONS:**

Adjunct Professor of Dermatology, China Medical University, Shenyang, China.

## **UNIVERSITY COMMITTEES**:

IRB July 1 – June 30, 2004

## KEHUA LI, MD

Assistant Professor, Department of Dermatology and Cutaneous Biology (continued)

# **PROFESSIONAL ORGANIZATIONS**:

- 1. Fellow of the American Academy of Dermatology.
- 2. Associate of the American Academy of Dermatopathology.
- 3. The Society for Investigative Dermatology.
- 4. American Society of Cosmetic Dermatology and Aesthetic Surgery.
- 5. American Medical Association.

## MŶ G. MAHONEY, PH.D.

Associate Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Brennan, D, Y Hu, A Kljuic, YW Choi, S Joubeh, JK Wahl, A Fertala L Pulkkinen J Uitto, AM Christiano, AA Panteleyev, and MG Mahoney. Differential structural properties and expression patterns suggest functional significance for multiple mouse desmoglein 1 isoforms. (Editorial invited submission to Differentiation).

Mishra SK, AH Talukder, AE Gururaj, Z Yang, RR Singh, MG Mahoney, C Franci, RK Vadlamudi, R Kumar. Upstream determinants of estrogen receptor-alpha regulation of metastatic tumor antigen 3 pathway. J Biol Chem. 2004 30;279(31):32709-15.

Kljuic A, H Bazzi, JP Sundberg, A Martinez-Mir, R O'Shaughnessy, MG Mahoney, M Levy, X Montagutelli, W Ahmad, VM Aita, D Gordon, J Uitto, D Whiting, J Ott, S Fischer, TC Gilliam, CA Jahoda, RJ Morris, AA Panteleyev, VT Nguyen, AM Christiano. (2003) Desmoglein 4 in hair follicle differentiation and epidermal adhesion: evidence from inherited hypotrichosis and acquired pemphigus vulgaris. Cell. 113(2):249-60.

Pulkkinen L, YW Choi, A Kljuic, J Uitto, MG Mahoney. (2003) Novel member of the mouse desmoglein gene family: Dsg1-beta. Exp Dermatol. 12(1):11-9.

## **GRANT SUPPORT**:

# "Molecular Characterization of Squamous Cell Carcinomas Derived from Recessive Dystrophic Epidermolysis Bullosa"

Principal Investigator: M.G. Mahoney, Ph.D.

Agency: Department of Defense, United States Army, 7/1/2002 – 6/30/2006

Direct cost: \$152,609/year

Elucidate the role of the metastsis associated protein (MTA1) in epithelial cell survival,

migration, and invasion.

## "Desmoglein 2 Function in Cell Adhesion and Pemphigus"

Principal Investigator: M.G. Mahoney, Ph.D.

Agency: National Institutes of Health, R03 Research Grant, 7/1/99 – 12/31/04

Direct Cost: \$50,000/year

Elucidate the role of the desmosomal adhesion molecule, desmoglein 2, in cellular proliferation, differentiation, and adhesion of epithelial tissues.

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

Christiano, A and MG Mahoney. The growing Family of Desmosomal Cadherins: And Then There Were Four. *Gordon Conference: Epithelial Differentiation and Keratinization*, Tilton, NH, July 2003.

Brennan, D, Y Hu, A Kljuic, JK Wahl, J Uitto, AA Panteleyev, AM Christiano, and MG Mahoney. "Complexity fo Dsg1- $\alpha$ , - $\beta$ , and - $\gamma$  expression in diverse mouse tissues and during hair follicle formation." *Society for Investigative Dermatology 2004*, Providence, RI. May 2004.

Nguyen, P, H Bazzi, A Kljuic, JH Vargas, M Dmochowski, M Barria, RF Ghohestani, LS Chan, J Uitto, LA Diaz, AM Christiano, MG Mahoney, and CT Nguyen. "Identification of a novel population of pathogenic autoantibodies against desmoglein 4 in different major variants of pemphigus." *Society for Investigative Dermatology 2004*, Providence, RI. May 2004.

Bazzi, H, A Kljuic, K Djabali, V Nguyen, Y Hu, D Brennan, MG Mahoney, and AM Christiano. "Desmoglein 4 is expressed in diverse tissue types and is localized to the desmosome." *Society for Investigative Dermatology* 2004, Providence, RI. May 2004.

## **HONORS/SPECIAL RECOGNITIONS:**

Review Panel: Reviewer, NIAMS-SDRC, National Institutes of Health, 2003

Scientific Review: Journal Clinical Investigation and Journal of Investigative Dermatology

## **PROFESSIONAL ORGANIZATIONS**:

Society for Investigative Dermatology The American Society for Cell Biology

Adjunct Associate Professor, Department of Dermatology and Cutaneous Biology

#### Main affiliation:

INSERM, Paris, France; Title: 1st class Research Director

#### **EDITORIAL ACTIVITIES**

Associate Editor: The Journal of Investigative Dermatology

Editorial Board Member: Experimental Dermatology

The Journal of Dermatological Science

Manuscript Reviews for:

American Journal of Pathology Archives of Dermatological Research

Arteriosclerosis, Thrombosis, and Vasc. Biology Biochemistry US

Biochimica Biophysica Acta Biochem Biophys Res Commun

British Journal of Dermatology British Journal of Cancer

British Journal of Pharmacology Cancer Research Circulation Research Current Genomics

EMBO Reports Experimental Cell Research
FASEB Journal Genes, Chromosomes and Cancer
Int. Journal of Biochemistry and Cell Biology International Journal of Cancer

Journal of Molecular Biology Laboratory Investigation

Médecine/Sciences Matrix Biology

Nature Genetics The Journal of Biological Chemistry
The Journal of Cell Science The Journal of Cellular Physiology

The Journal of Clinical Investigation The Journal of Immunology

The Journal of International Dermatology JNCI

Trends in Molecular Medicine

#### **HONORS:**

Chairman, session "Vascular Biology", Annual Meeting of the European Society for Dermatological Research, Vienne, Autriche, Septembre 2004

Co-chairman, Cellular signaling session, VIIIth Inernational Scleroderma Workshop. Cambridge, U.K., 2004

Contrat d'Interface INSERM-AP-HP, 2003

Chairman, session "Fibroblasts – Effectors and Regulators of Fibrosis", Scleroderma Spectrum Disorders Post-graduate Course 2003, Royal Free Hospital, London UK, Février 2003

- "Ad Hoc" Reviewer, "Gene Expression and Cellular Signaling" abstracts, International Investigative Dermatology Meeting, Miami, FL, 2003
- "Ad Hoc" Reviewer d'un "Program Project" pour la British Heart Foundation, 2003
- "Ad Hoc" Grant Reviewer for the Binational Science Foundation, USA-Israel, 2004
- "Ad Hoc" Reviewer, "Cellular Signalling" abstracts, Annual Meeting of the European Society for Dermatological Research, Vienna, Austria, 2004

Adjunct Associate Professor, Department of Dermatology and Cutaneous Biology (continued)

#### **PEER-REVIEWED PUBLICATIONS:**

Brellier, F., Marionnet, C., Chevalier-Lagente, O., Toftgards, R., <u>Mauviel, A.</u>, Sarasin, A., and Magnaldo, T. Ultraviolet irradiation regulates *PATCHED* gene transcription in human epidermal keratinocytes through an AP-1-dependent process. *Cancer Res.*, 2004, 64:2699-2704

Xavier, S., Piek, E., Fuji, M., Javelaud, D., <u>Mauviel, A.</u>, Samuni, A., Flanders, K.C., Reiss, M., Yarkoni, S., Mitchell, J.B., Roberts, A.B., and Russo, A. Amelioration of Radiation-Induced Fibrosis: Down regulation of TGF-b Signaling by halofuginone. *J. Biol. Chem.*, 2004, 279:15167-15176

Verrecchia, F., Tacheau, C., Wagner, E.F., and <u>Mauviel, A</u>. A Central Role for Jun-N-Terminal Kinase Activity in Mediating the Antagonistic Activity of Pro-inflammatory Cytokines Against Transforming Growth Factor-β-Induced Smad3/4 Signaling. *J. Biol. Chem.*, 2003, <u>278</u>:1585-1593

Higashi, K., Inagaki, Y., Suzuki, N., Mitsui, S., <u>Mauviel, A.</u>, Kaneko, H., and Nakatsuka, I. Ybox binding protein YB-1 mediates transcriptional repression of human a2(I) collagen gene expression by interferon-g. *J. Biol. Chem.*, 2003, 278:5156-5162

Javelaud, D., Laboureau, J., Gabison, E., Verrecchia, F., and <u>Mauviel, A</u>. Disruption of Basal Jun-N-Terminal Kinase (JNK) Activity Differentially Affects Key Fibroblasts Functions Important for Wound Healing. *J. Biol. Chem.* 2003, <u>278</u>:24624-24628

Gabison, E., Chastang, P., Menashi, S., Oster, M., <u>Mauviel, A.</u>, and Hoang-Xuan, T. Late Corneal Perforation Following Photorefractive Keratectomy Associated with Topical Diclofenac: Involvement of Matrix Metalloproteinases. *Ophtalmology*, 2003, 110:1626-1631

Wendling, J., Marchand, A., <u>Mauviel, A.</u>, and Verrecchia, F. 5-fluoro-uracyl antogonizes TGF-β/Smad signaling and transcriptionally represses *COL1A2* gene expression. Implication for the treatment of fibrotic disorders. *Mol. Pharmacol.*, 2003, 64:707-713

Laurent, M., Martinerie, C., Thibout, H., Verrecchia, F., Hoffman M,P, Le Bouc, Y., <u>Mauviel, A.</u>, and Kleinman, H. NOV H induces the expression of mmp3 gene in glioblastoma cells and enhances their migration. *FASEB J.* 2003, 17:1919-1921

Pendaries, V., Verrecchia, F., Michel, S., and <u>Mauviel, A</u>. Retinoic Acid Receptors Interfere with the TGF-β/Smad Signaling Pathway in a Ligand-Specific Manner. *Oncogene*, <u>22</u>:8212-8220 Schiller, M., Verrecchia, F. and <u>Mauviel, A</u>. Cyclic Adenosine 3',5'-Monophosphate-Elevating Agents Inhibit Transforming Growth Factor-β-Induced SMAD3/4-Dependent Transcription Via a Protein Kinase A-Dependent Mechanism. *Oncogene*, <u>22</u>:8881-8890

Adjunct Associate Professor, Department of Dermatology and Cutaneous Biology (continued)

Marionnet, C., Dumas, A., Verrecchia, F., Mollier, K., Compan, D., Bernard, B., Bernerd, F., Lahfa, M., Leclaire, J., Medaisko, C., Mehul, B., Seité, S., <u>Mauviel, A.</u>, and Dubertret, L. Microarray-based analysis of stress-induced genes in human epidermis *in vivo. J. Invest. Dermatol.*, 121:1447-1458

## **REVIEW ARTICLES AND BOOK CHAPTERS:**

Javelaud, D. et <u>Mauviel</u>, <u>A</u>. Transforming Growth Factor-bs: signalisation et rôles physio-pathologiques. *Pathol. Biol.*, 2004, <u>52</u>:50-54

Verrecchia, F., and <u>Mauviel</u>, <u>A</u>. Control of type I collagen gene expression by TGF-β, interferences with pro-inflammatory cytokines. *Cell. Signal.*, 2004, <u>16</u>:873-880

Javelaud, D. and <u>Mauviel</u>, <u>A</u>. Molecule in Focus: Mammalian TGF-βs, Mammalian Transforming growth factor-βs: Smad signaling and physio-pathological roles *Int. J. Biochem. Cell Biol.* 2004, 36:1161-1165

Schiller, M., Javelaud, D., and Mauviel, A. TGF- $\beta$ , a masterswitch for tissue remodeling: signaling via the Smad pathway. *J. Dermatol. Sci.*, 2004, <u>35</u>:83-92

Javelaud, D., Verrecchia, F. et <u>Mauviel</u>, <u>A</u>. TGF-β et contrôle des fonctions fibroblastiques impliquées dans la réparation tissulaire. *Cours de Biologie de la Peau*. (C. dezutter-dambuyant Ed.), INSERM editions scientifiques, 2004, pp.137-149

Mauviel, A. Transforming growth factor-β: the Smad pathway and its implications in fibrosis. In *Methods in Molecular Medicine*. Varga, Phan and Brenner Eds, Humana Press, sous presse

Gabison, E., Hoang-Xuang, T., <u>Mauviel, A.</u>, and Menashi, S. Métalloprotéinases et angiogenèse. *Pathol. Biol.* 2003, <u>51</u>:161-166

## **INVITED PRESENTATIONS:**

Cytokines to Antagonize TGF-beta/Smad Signaling: Implications for Wound Healing. Serono Pharmaceutical Research Institute, Genève, Suisse, Février 2004

Interférences Transcriptionnelles de la voie Jun N-terminal Kinase/AP-1 avec la machinerie Smad en aval du TGF-beta.

Institut Paoli-Calmettes, IFR 57 "Institut de Cancérologie et d'Immunologie de Marseille, Marseille, Avril 2004

Interférences Transcriptionnelles de la Voie AP-1 avec la Machinerie Smad en Aval du TGFbeta. Implication pour le Remodelage de la Matrice Extracellulaire. Institut Curie, Orsay, Mai 2004

Adjunct Associate Professor, Department of Dermatology and Cutaneous Biology (continued)

Transcriptional control of fibrillar collagen genes by TGF-beta and inflammatory cytokines XIXth FECTS Meeting, Taormina, Italie, Juillet 2004

The Regulation of Fibroblast Functions by JNK 8th International Workshop On Scleroderma Research, Cambridge, U.K., Août, 2004

Antagonizing TGF-β Signaling: a Key Role for the JNK Pathway Celgene Corp., San Diego, CA, Janvier 2003

AP-1 interference with the TGF-β/Smad Pathways: Implications for ECM remodeling 1/ Scleroderma Spectrum Disorders Post-graduate Course, Royal Free Hospital Londres, UK, Février 2003 (invité par C. Black)

2/ LondonMatrix Group Monthly Meeting, UCL, Londres, Février 2003

TGF-β et matrice conjonctive COBIP (Cours de Biologie de la Peau), CIRC, Lyon, Mars 2003

Smad signaling and fibrosis

Gordon Research Conference on Tissue Repair, Il Coccio, Barga, Italie, Juin 2003

Interactions transcriptionnelles entre le TGF-beta et les cytokines pro-inflammatoires: rôle de la voie Jun N terminal kinase

Symposium "TGF-Beta: Voies de transduction et Rôle Physiologique", IFR 14, Jussieu, Paris, Juin 2003

Antagonizing TGF-β/Smad Signaling: a Key Role for the c-Jun N-terminal Kinase Pathway FASEB Summer Research Conference "The TGF-β Superfamily: Signaling and Development". Tucson, AZ, Juillet 2003

Molecular mechanisms underlying AP-1 antagonistic activity against TGF-β/Smad signaling Research Institute of Molecular Pathology (I.M.P.), Vienna, Autriche, Août 2003

JNK as a critical effector of TNF antagonism against TGF- $\beta$ /Smad signaling. University hospital, Aachen, Allemagne, Décembre 2003

#### **MEMBERSHIPS TO SCIENTIFIC SOCIETIES:**

Society for Investigative Dermatology American Society for Biochemistry and Molecular Biology European Society for Dermatologic Research Wound Healing Society French Society for Connective Tissue (Board Member)

#### JOSEPH L. PACE MD FRCP EDIN FRCP LOND FCPP FAAD KM

Adjunct Clinical Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Clinical significance of malignant Melanoma precursors in Cutaneous Melanoma Giuseppe Noto Ed Avantgarde Publ pp 23-39 Dec 2002

Characteristics of superficial mycoses in the Maltese islands - co-author Int J Dermatol 2003, 42, 265-271

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

Chair 4<sup>th</sup> Maltese Medical School Conference December 2003 **Mediterranean Leishmaniasis** Philadelphia College of Physicians Feb 2004 The **Medical Heritage of a Maltese dermatologist**-History Society Washington AAD Feb 2004

Adult acne AMED Congress Montpellier June 2004

Acne-the rise and fall of antibiotics First Libyan Derm Congress Benghazi June 2004

## **HONORS/SPECIAL RECOGNITIONS:**

Knight of the Sovereign and Military Order of Malta, Rhodes, and Jerusalem

Board Member European Academy of Dermatology & Venereology

Board member, AMED Association of Mediterranean dermatologists

Editorial Board Member Journal of the European Academy of Dermatology & Venereology President Maltese Dermatology Association

Adjunct Clinical Professor of Dermatology, Jefferson Medical College of Thomas Jefferson University, Philadelphia

Senior Lecturer Dept. of Medicine (Dermatology), University of Malta Medical School

Examiner in Medicine (Final Year) & Dermatology (4th Year) Faculty of Medicine,

University of Malta

Chairman Dermatology/Rheumatology Research Group, Faculty of Medicine, University of Malta

Member Postgraduate Committee, Faculty of Medicine, University of Malta

Vice President Europe International Academy of Cosmetic Dermatology

President Rotary Club Malta

## PROFESSIONAL ORGANIZATIONS:

Maltese Association of Dermatology and Venereology (President)

European Academy of Dermatology & Venereology (Secretary-General)

Mediterranean Association of Dermatology (Vice-President)

American Academy of Dermatology

International Academy of Cosmetic Dermatology (Vice President Europe)

International Society of Dermatology

#### LAWRENCE CHARLES PARISH, MD

Clinical Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Parish LC, Oumeish OY, Millikan LE, Kurban A. Dubai Derma 2003: Dubai World Dermatology and Laser Conference and Exhibition. May 11-13, 2003, Dubai, United Arab Emirates. SKINmed. 2003 Jul-Aug;2(4):256-7.

Wolf R, Parish LC. Who's the boss? Clin Dermatol. 2003 Jul-Aug;21(4):335-6.

Oumeish OY, Wolf R, Parish LC. Risks and precautions of chemical warfare agents. SKINmed. 2003 Jul-Aug;2(4):205.

Oumeish OY, Parish LC. Background, problems, and perspectives of management of common pediatric skin problems in developing countries. Clin Dermatol. 2003 Jul-Aug;21(4):254-9.

Witkowski JA, Parish LC. Antimicrobials in the treatment of acne and rosacea. SKINmed. 2003 Jul-Aug;2(4):202.

Parish LC, Witkowski JA. Does complementary medicine work? Dermatol Ther. 2003;16(2):85-6.

Parish LC. Parish JL. Cosmetic dermatology and Art Deco: lookin' good. SKINmed. 2003 Sep-Oct; 2(5): 270-1.

Parish LC. SKINmed achieves *Index Medicus* status. SKINmed. 2003 Nov-Dec; 2(6): 333-4.

Wolf R, Orion E, Parish LC. A scientific soap opera and winter itch. SKINmed. 2004 Jan-Feb; 3(1): 9-10.

Parish LC, Witkowski JA. Controversies about the decubitus ulcer.Dermatol Clin. 2004 Jan; 22(1): 87-91.

Parish LC, Witkowski JA. The bedbugs never left. SKINmed. 2004 Mar-Apr; 3(2): 69-70.

Wolf R, Matz H, Parish LC. After the antibiotics and breast cancer study: what to tell patients. SKINmed. 2004 May-Jun; 3(3): 125-7.

Witkowski JA, Parish JL, Parish LC. Traumatic purpuric penile ulcer. Acta Dermatovenerol Croat. 2004; 12(2): 96-8.

#### LAWRENCE CHARLES PARISH, MD

Clinical Professor, Department of Dermatology and Cutaneous Biology (continued)

## **LAY PUBLICATIONS:**

Parish LC. Pyogenic granuloma. Pediatrics for Parents. 2003; 20(12): 5.

Parish LC. Dyshidrosis. Pediatrics for Parents. 2004; 21 (1): 12.

## **POSTERS:**

Wolf R, Orion E, Parish LC. Who's the boss? Israel Society of Dermatology and Venereology, Eilat, Israel, June 17-18, 2004

## **PRESENTATIONS:**

Evidence-based care of decubitus ulcers. American Academy of Dermatology, Washington, DC. February 7, 2004

The other medicine: alternative, complementary, and integrative. Dubai Derma 2004, Dubai, United Arab Emirates, April 27, 2004.

The decubitus ulcer in perspective. Dubai Derma 2004, Dubai, United Arab Emirates, April 29, 2004.

The controversial wound known as the decubitus ulcer. Israel Society of Dermatology and Venereology, Eilat, Israel, June 17, 2004

## **SEMINARS ORGANIZED AND CHAIRED:**

History of Dermatology Seminar, History of Dermatology Society, Washington, DC, February 5. 2004.

Integrative Dermatology & the Clinician. American Academy of Dermatology, Washington, DC. February 6, 2004

#### LAWRENCE CHARLES PARISH, MD

Clinical Professor, Department of Dermatology and Cutaneous Biology (continued)

## **MEETINGS ATTENDED:**

64<sup>th</sup> Annual Meeting of the American Academy of Dermatology, Washington, DC. February 6-11, 2004

Dubai Derma 2004, Dubai, United Arab Emirates, April 27 -29, 2004.

28<sup>th</sup> Annual Meeting of the Israel Society of Dermatology and Venereology, Eilat, Israel, June 17-18, 2004

## PROFESSIONAL ORGANIZATIONS:

## **Member or Fellow:**

American Association for the History of Medicine, American College of Physicians, American Dermatological Association, American Academy of Dermatology, Canadian Dermatology Association, Canadian Society for the History of Medicine, College of Physicians of Philadelphia (Section on the History of Medicine of the College of Physicians of Philadelphia, Section on Dermatology of the College of Physicians of Philadelphia), European Academy of Dermatology and Venereology. European Society for the History of Dermatology and Venereology, French Society for the History of Dermatology and Venereology, History of Dermatology Society (president), International Academy of Cosmetic Dermatology (president), Pennsylvania Academy of Dermatology, Philadelphia Dermatological Society

## **Honorary Member or Fellow:**

American College of Veterinary Dermatology, Bulgarian Society of Dermatology, Florida Society of Dermatology, Italian Society of Dermatology and Venereology, Polish Academy of Dermatology, Venezuelan Society of Dermatology,

#### **Corresponding Member or Fellow:**

British Association of Dermatologists, French Society of Dermatology and Syphilolography, Israeli Society of Dermatology

#### ELLEN G. PFENDNER, PHD

Assistant Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Sprecher E, Yossipovitch G, Bergman R, Ciubutaro D, Indelman M, Pfendner E, Goh CL, Miller CJ, Uitto J, Richard G 2003 Epidermolytic hyperkeratosis and epidermolysis bullosa simplex caused by frameshift mutations altering the V2 tail domains of keratin 1 and keratin 5. *J Invest Dermatol* 120:623-626.

Ciubutaro D, Bergman R, Baty D, Indelman M, Pfendner E, Petronius D, Moualem H, Kanaan M, Ben Amitai D, McLean WHI, Uitto J, Sprecher E 2003 Epidermolysis bullosa simplex in Israel: clinical and genetic features. Arch Dermatol 139:498-505.

Uitto, J, E Pfendner, L G Jackson Probing the fetal genome: progress in non-invasive prenatal diagnosis. 2003 Trends in Molecular Medicine 9:339-343.

Schara U, Tucke J, Mortier W, Nusslein T, Rouan F, Pfendner E, Zillikens D, Bruckner-Tuderman L, Uitto J, Wiche G, Schroder R. 2004 Severe mucous membrane involvement in epidermolysis bullosa simplex with muscular dystrophy due to a novel plectin gene mutation. European Journal of Pediatrics 163:218-22.

Presentations given at the Society of Investigative Dermatology Meeting, April 27-May 1 2004 Providence Rhode Island:

Pfendner, E, Uitto, J 2004 Novel and recurrent mutations in the keratin 5 and 14 genes in epidermolysis bullosa simplex: implications for genetic counseling. Journal of Investigative Dermatology 123 in press

Cash, S, Sadowski, S, Uitto, J, Pfendner, E 2004 Recurrent and de novo dominant COL7A1 mutations in dystrophic forms of epidermolysis bullosa. Journal of Investigative Dermatology 123 in press

Sadowski, S,Uitto, J, Pfendner, E, 2004 Recurrent ITGB4 mutations in Hispanic patients with epidermolysis bullosa and pyloric atresia. Journal of Investigative Dermatology 123 in press

Uitto, J, Pfendner, E, 2004 Compound heterozygosity for unique in-frame insertion and deletion mutations in the plectin gene in a mild case of epidermolysis bullosa with very late onset muscular dystrophy. Journal of Investigative Dermatology 123 in press.

Fratta, S, Uitto, J, Terry, S, Terry, P, Ringpfeil, F, Pfendner, E, 2004 dHPLC screening detects novel and recurrent mutations in Pseudoxanthoma Elasticum. Journal of Investigative Dermatology 123 in press.

## ELLEN G. PFENDNER, PHD

Assistant Professor, Department of Dermatology and Cutaneous Biology (continued)

# Presentation at DebRA National Patient Conference, March 25-28 2004 Palo Alto CA:

Genetics and Genetic Testing for Epidermolysis Bullosa

# **UNIVERSITY COMMITTEES:**

May 2003 to June 2004 Member of IRB Thomas Jefferson University and Jefferson Medical College Reviewer of proposal submitted for IRB approval for clinical trial and research involving human subjects.

## **MEMBERSHIPS:**

American Society of Human Genetics Society of Investigative Dermatology

Associate Professor, Department of Dermatology and Cutaneous Biology Associate Professor, Department of Medicine

## **PUBLICATIONS:**

- F. Rouan, C.W. Lo, A. Fertala, M. Wahl, U. Rodeck, J. Uitto, <u>G. Richard</u>. Divergent effects of two sequence variants of *GJB3* (G12D and R32W) on function of connexin 31 in vitro. <u>Exp Dermatol</u> 12: 191-197, 2003
- V. Lagrée, K. Brunschwig, <u>G. Richard</u>, M. M. Falk, and N. B. Gilula. Dominant Negative Inhibition Of Connexin-43 And Connexin-32 By Connexin-43 Substitution Variants: Implications For The Role Of Specific Amino Acid Residues In Connexin Function And Selectivity. <u>J Cell Sci</u> 116: 3189-3201, 2003.
- P. H. Itin, M. Moschopulos, G. Richard. Reticular erythrokeratoderma (Aara disease) A new disorder of cornification. Am J Med Genet 120A:237-240, 2003
- E. Sprecher, G. Yossipovitch, R. Bergman, D. Ciubutaro, M. Indelman, E. Pfender, L. C. Goh, Ch. J. Miller, J. Uitto, <u>G. Richard</u>. Epidermolytic hyperkeratosis and epidermolysis bullosa simplex caused by frameshift mutations altering the V2 tail domains of keratin 1 and keratin 5. <u>J. Invest Dermatol</u> 120: 623-6, 2003
- Ishida-Yamamoto, <u>G. Richard</u>, H. Takahasi, H. Iizuka. *In vivo* studies of mutant K1 in Ichthyosis Hystrix Curth-Macklin. <u>J Invest Dermatol</u> 120: 498-500, 2003
- L.Y. Hwang, J.B. Lee, <u>G. Richard</u>, J. Uitto, S. Hsu. Type I segmental manifestation of Hailey-Hailey disease. <u>J Am Acad Dermatol</u> 49 (4): 712-714, 2003
- Indelman M, Hamel CP, Bergman R, Nischal KK, Thompson D, Surget MO, Ramon M, Ganthos H, Miller B, <u>Richard G</u>, Lurie R, Leibu R, Russell-Eggitt I, Sprecher E. Phenotypic diversity and mutation spectrum in hypotrichosis with juvenile macular dystrophy. <u>J Invest Dermatol</u> 121(5):1217-20, 2003
- <u>G. Richard</u>, N. Brown, F. Rouan, J.-G. Van der Schroeff, E. Bijlsma, L. F. Eichenfield, V. P. Sybert, K. E. Greer, P. Hogan, C. Campanelli, J. G. Compton, S. J. Bale, J. J. DiGiovanna, J. Uitto. Genetic heterogeneity in erythrokeratodermia variabilis: Novel mutations in the connexin gene *GJB4* (Cx30.3) and genotype-phenotype correlations. <u>J Invest Dermatol</u> 120:601-609, 2003
- W. Brown, M. L. Levy, C. M. Flaitz, B. S. Reid, S. Manolidis, A. A. Hebert, M. M. Bender, H. A. Heilstedt, K. S. Plunkett, P. Fang, P. Chung, B. B. Roa, H.-Y. Tang, G. <u>Richard</u>, R. L. Alford. A novel *GJB2* (Cx26) mutation, F142L, in a patient with unusual mucocutaneous findings and deafness. J Invest Dermatol 121(5): 1221-1223, 2003

Associate Professor, Department of Dermatology and Cutaneous Biology Associate Professor, Department of Medicine (continued)

- A.Y. Jan, S. Amin, P. Ratajczak, <u>G. Richard</u>, V.P. Sybert. Genetic heterogeneity of KID syndrome: Identification of a Cx30 gene (*GJB6*) mutation in a patient with KID syndrome and congenital atrichia. <u>J Invest Dermatol</u> 122:1108-1113, 2004.
- Topaz, D.L. Shurman, R. Bergman, M. Indelman, P. Ratajczak, M. Mizrachi, Z. Khamaysi, D. Behar, D. Petronius, V. Friedman, I. Zelikovic, S. Raimer, A. Metzker, <u>G. Richard</u>, E. Sprecher. Mutations in *GALNT3*, encoding a protein involved in O-linked glycosylation, cause familial tumoral calcinosis. <u>Nat Genet</u>, 36:579-81, 2004; epub May 09, 2004.
- <u>Richard</u>, N. Brown, A. Ishida-Yamamoto and A. Krol. Expanding the phenotypic spectrum of Cx26 disorders: Bart-Pumphrey Syndrome is caused by a novel missense mutation in *GJB2*. <u>J Invest Dermatol</u>, 2004, in press
- Sprecher, E, Tesfaye-Kedjela, A, Ratajczak, P, Bergman, R, <u>Richard-G</u>: Deleterious mutations in SPINK5 in a patient with congenital ichthyosiform erythroderma: molecular testing as a helpful diagnostic tool for Netherton syndrome. <u>Clin Exp Dermat</u>, 2004, in press
- Ishida-Yamamoto, C. Deraison, C. Bonnart, E. Bitoun, T.J. O'Brien, K. Wakamatsu, S. Ootsubo, H. Takahashi, Y. Hashimoto, P. JC Dopping-Hepenstal, J. A. McGrath, H. Iizuka, <u>G. Richard</u>, A. Hovnanian: vTransport of LEKTI via lamellar granules: relevance to desquamation in normal human skin and the pathogenesis of Netherton syndrome. <u>J Invest Dermatol</u>, submitted 6/2004
- A.S. Geyer, P. Ratajczak, M. Pol-Rodriguez, M. Garzon, G. Richard. Netherton syndrome with extensive skin peeling and failure to thrive due to a homozygous frameshift mutation in *SPINK*. Br. J Dermatol, submitted June 2004

#### **INVITED REVIEWS:**

- G. Richard. Connexin gene pathology. Clin Exp Dermatol 28 (4):397-409, 2003
- <u>G. Richard.</u> Molecular genetics of the ichthyoses. In: Genetic Disorders of the Skin. Am J Med Genet (Seminars), Der Kaloustian and Happle (eds), in press
- J. Uitto and <u>G. Richard</u>. Progress in epidermolysis bullosa: Genetic classification and clinical implications. In: Genetic Disorders of the Skin. Am J Med Genet (Seminars), Der Kaloustian and Happle (eds), in press

#### OTHER PUBLICATIONS AND EDITORIALS:

J. Uitto and <u>G. Richard</u>. Heritable skin disorders – Novel mutations and spectrum of phenotypes. Editorial. J Invest Dermatol 121:V, 2003

Associate Professor, Department of Dermatology and Cutaneous Biology Associate Professor, Department of Medicine (continued)

## **BOOK CHAPTERS:**

- <u>G. Richard</u>. Erythrokeratodermia variabilis. In: The NORD Guide to Rare Diseases. Lippincott, Williams & Wilkins, 2003.
- <u>G. Richard</u>. Progressive symmetric erythrokeratodermia. In: The NORD Guide to Rare Diseases. Lippincott, Williams & Wilkins, 2003.
- <u>G. Richard</u> and J. Uitto. Inherited Skin disease. In: Enzyclopedic Reference of Genomics and Proteomics in Molecular Medicine. D. Ganten & K. Ruckpaul (eds) Springer International, in press.
- J. Uitto, <u>G. Richard</u> and A. Christiano. Epidermolysis Bullosa. In: Principles of Molecular Medicine, in press

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

## **Oral presentations**

International gap Junction Conference, Cambridge, UK. August 23-28, 2003. Molecular Studies in KID Syndrome Reveal a Mutational 'Hot Spot' in Cx26, Genetic Heterogeneity and an Intriguing Phenotypic Variability. <u>G. Richard</u>, L. Yi, D. Wasserman, F. Rouan, A.Y. Jan, V.P. Sybert, J. Uitto.

Invited lecture, Wistar Institute, Philadelphia, PA (Melanoma Group Seminar), October 28, 2003. 'Connecting with Connexins'. G. Richard

Annual Meeting of the Assoc. for Research in Vision and Ophthalmology, Ft. Lauderdale, April 25-30, 2004. Connexin expression in the human corneal epithelium in vivo and in vitro. D.I. Shurman, L. Glazewski, J.D. Zieske and <u>G. Richard</u>

Annual Meeting of the American Academy of Dermatology. February 2004. Co-organizer and Faculty, Forum 'Impact of Molecular Genetics on Clinical Dermatology and Dermatopathology'. Clinical Implications of Advances in Molecular Genetics of DOC.

Annual Meeting of the Society of Pediatric Dermatology. February 2004. Erythrokeratodermia variabilis in a Hispanic child. AB Alio, <u>G Richard</u>, MA Dohil, C Maari, LF Eichenfield.

Associate Professor, Department of Dermatology and Cutaneous Biology Associate Professor, Department of Medicine (continued)

Annual Meeting of the Assoc. for Research in Vision and Ophthalmology, Ft. Lauderdale, April 25-30, 2004. Connexin expression in the human corneal epithelium in vivo and in vitro. D.I. Shurman, L. Glazewski, J.D. Zieske and <u>G. Richard</u>

Summer Meeting of the American Academy of Dermatology. July 2004. Invited speaker 'Connexins in Pediatric Dermatoses'.

Invited lecture. Grand Rounds, Department of Dermatology, NorthWestern University, Chicago. May 26, 2004. Human Connexin Disorders: Mind the Gap Junction

Invited lecture. Cell Biology Interest Group Meeting, NorthWestern University, Chicago. May 26, 2004. Connexin mutations and their functional consequences in ectodermal epithelia.

## ABSTRACTS AND POSTER PRESENTATIONS:

Gordon Research Conference. Molecular and Functional Studies in KID Syndrome Reveal Genetic Heterogeneity and Functional Impairment of Cx26. <u>G. Richard</u>, L. Yi, D. Wasserman, F. Rouan, A.Y. Jan, V.P. Sybert, J. Uitto.

International gap Junction Conference, Cambridge, August 23-28, 2003. Similar Mutations of Cx31 and Cx30.3 Impair Gap Junction Function, Dominantly Inhibit wtCx31, and Have Different Consequences for Cell Survival. F. Rouan, L. Yi, J. Uitto, <u>G. Richard</u>.

Sigma Xi Research Day, Jefferson Medical College, March 18, 2004. Calcium-Induced Changes in Connexin Gene Expression in Corneal Epithelial Cell Cultures. LM Glazewski, DL Shurman and <u>G Richard</u>.

Sigma Xi Research Day, Jefferson Medical College, March 18, 2004. Linkage studies in the skin disorder Pityriasis Rubra Pilaris exclude four prominent candidate gene regions. K. Richard, F. Ringpfeil, and <u>G. Richard</u>.

Annual Meeting of the American Society of Medical Genetics. *In vitro* functional analysis of a Connexin 26 protein bearing an F142L amino acid substitution. Hsiao-Yuan Tang, Chester W. Brown, <u>Gabriele Richard</u>, Raye Lynn Alford.

65th Annual Meeting of The Society for Investigative Dermatology, April 28-May2, 2004. Molecular Epidemiology Of Epidermolysis Bullosa In Middle East Populations. E. Sprecher, J. Abu Sa'd, E. Pfendner, M. Indelman, D. Ciubutaro, M. Mizrachi, G.G. Lestringant, L. Pulkkinen, G. Richard, M. Kanaan, J. Uitto, R. Bergman.

Associate Professor, Department of Dermatology and Cutaneous Biology Associate Professor, Department of Medicine (continued)

65th Annual Meeting of The Society for Investigative Dermatology, April 28-May2, 2004. Netherton Syndrome: Novel and Recurrent Mutations in SPINK5 and implications for screening and diagnosis. <u>Gabriele Richard</u>, Paulina Ratajczak, Shivan Amin, Humza Ilyas, Aida Tesfaye Kedjela Elaine C. Siegfried and Jouni Uitto.

65th Annual Meeting of The Society for Investigative Dermatology, April 28-May2, 2004. A novel dominant missense mutation of GJB6 (Cx30) causing KID syndrome with mild cutaneous phenotype. D. Wasserman, P. Ratajczak, F. Ringpfeil, G. Richard.

65th Annual Meeting of The Society for Investigative Dermatology, April 28-May 2, 2004. Calcium-Induced Changes in Connexin Expression and Function in the Corneal Epithelium. D.I. Shurman, L. Glazewski, J.D. Zieske and G. Richard.

## **DEPARTMENTAL COMMITTEES**

Co-Director, Molecular Diagnostic Laboratory

Member of the Organizing Committee for the 12th International Symposium on Basement Membranes, Philadelphia, June 15 - 18, 2005

## **PROFESSIONAL ORGANIZATIONS**:

American Society of Human Genetics Society of Investigative Dermatology Association for Research in Vision and Ophthalmology German Society of Dermatology Society of Pediatric Dermatology Philadelphia Dermatological Society Medical Advisory Board, FIRST

#### FRANZISKA RINGPFEIL, MD

Assistant Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Uitto J, <u>Ringpfeil</u> F. Ehlers-Danlos syndrome-molecular genetics beyond the collagens. J Invest Dermatol. 122:xii-xiii, 2004

<u>Ringpfeil</u> F, McGuigan K, Kozic H, Uitto J. Pseudodominance in Pseudoxanthoma Elasticum - a common Phenomenon: Implications for Genetic Counseling. (submitted)

## **BOOK CHAPTERS:**

Uitto J, <u>Ringpfeil</u> F. Heritable Disorders of Connective Tissue. In: Principles of Molecular Medicine (2<sup>nd</sup> Edition). Diaz LA, Goldsmith LA, The Humana Press, Totowa, NJ (in press)

<u>Ringpfeil</u> F, Uitto J. Pseudoxanthoma Elasticum. In: Encyclopedia of Diagnostic Genomics and Proteomics. Fuchs J, Podda M, Marcel Dekker, New York, NY, (in press)

#### **ABSTRACTS:**

<u>Ringpfeil</u> F, McGuigan K, Kozic H, Uitto J. Pseudodominance in Pseudoxanthoma Elasticum - a common Phenomenon: Implications for Genetic Counseling. American Society for Human Genetics, Fifty-Third Annual Meeting, Los Angeles, CA, November 4-9, 2003

Fuchsel L, Kozic H, McGuigan K, Skvarka C, Jacobson M, Uitto J, <u>Ringpfeil</u> F. Genotype Phenotype Correlation in 62 Patients with Pseudoxanthoma Elasticum. Society for Investigative Dermatology, Providence, RI, April 28-May 1, 2004 (J Invest Derm 122:A93)

<u>Ringpfeil</u> F., McGuigan K, Fuchsel L, Kozic H, Lebwohl M, Uitto J. Autosomal Dominant Inheritance in PXE revisited. Society for Investigative Dermatology, Providence, RI, April 28-May 1, 2004 (J Invest Derm 122:A92)

Wasserman DI, Ratajczak P, <u>Ringpfeil</u> F, Richard G. A novel dominant missense Mutation of GJB6 (Cx30) causing KID Syndrome with mild cutaneous Phenotype. Society for Investigative Dermatology, Providence, RI, April 28-May 1, 2004 (J Invest Derm 122:A92)

Rucker D, Campanelli C, Lee J, <u>Ringpfeil</u> F. Widespread Vitiligo in an Infant. Society for Pediatric Dermatology Annual Meeting, Charleston, SC, June 17-20, 2004

#### **GRANT SUPPORT:**

Dermatology Foundation; Clinical Career Development Award

#### FRANZISKA RINGPFEIL, MD

Assistant Professor, Department of Dermatology and Cutaneous Biology (continued)

## PARTICIPATION IN SCIENTIFIC/ CLINICAL MEETINGS AND PRESENTATIONS:

<u>Ringpfeil</u> F, McGuigan K, Kozic H, Uitto J. Pseudodominance in Pseudoxanthoma Elasticum - a common Phenomenon: Implications for Genetic Counseling, American Society for Human Genetics, Fifty-Third Annual Meeting, Los Angeles, CA, November 4-9, 2003

Society for Pediatric Dermatology, 16<sup>th</sup> Annual Pre-AAD Meeting, Washington DC, February 5, 2004

Amir I, <u>Ringpfeil</u> F. Sweet Syndrome with Hemophilus Influenza Pneumonia, American Academy of Dermatology, 62<sup>nd</sup> Annual Meeting, Washington, D.C. February 6-11, 2004

Cenci J, <u>Ringpfeil</u> F. Bullous Lichen Planus in a Child, American Academy of Dermatology, 62<sup>nd</sup> Annual Meeting, Washington, D.C. February 6-11, 2004

<u>Ringpfeil</u> F. Genotype Phenotype Correlation in Pseudoxanthoma Elasticum. Plenary Session, Clinical Scholars Program. Society for Investigative Dermatology, Providence RI, April 28- May 1, 2004

Fuchsel L, Kozic H, McGuigan K, Skvarka C, Jacobson M, Uitto J, <u>Ringpfeil</u> F. Genotype Phenotype Correlation in 62 Patients with Pseudoxanthoma Elasticum. Society for Investigative Dermatology, Providence, RI, April 28-May 1, 2004

<u>Ringpfeil</u> F., McGuigan K, Fuchsel L, Kozic H, Lebwohl M, Uitto J. Autosomal Dominant Inheritance in PXE revisited. Society for Investigative Dermatology, Providence, RI, April 28-May 1, 2004

Wasserman DI, Ratajczak P, <u>Ringpfeil</u> F, Richard G. A novel dominant missense Mutation of GJB6 (Cx30) causing KID Syndrome with mild cutaneous Phenotype. Society for Investigative Dermatology, Providence, RI, April 28-May 1, 2004

Rucker D, Campanelli C, Lee J, <u>Ringpfeil</u> F. Widespread Vitiligo in an Infant. Society for Pediatric Dermatology Annual Meeting, Charleston, SC, June 17-20, 2004

#### **University Committees:**

JUP Advisory Council

## FRANZISKA RINGPFEIL, MD

Assistant Professor, Department of Dermatology and Cutaneous Biology (continued)

# **MEMBERSHIP IN PROFESSIONAL ORGANIZATIONS:**

- American Academy of Dermatology
- Dermatology Foundation, Vice Chair, Pennsylvania
- Society for Investigative Dermatology
- Society for Pediatric Dermatology
- Philadelphia Dermatological Society
- Medical Advisory Board, NAPE (National Association for Pseudoxanthoma elasticum)
- Airline Ambassadors International

#### FATIMA ROUAN, PH.D.

Research Assistant Professor, Department of Dermatology and Cutaneous Biology

# **PUBLICATIONS:**

Schara U, Tucke J, Mortier W, Nusslein T, **Rouan F**, Pfendner E, Zillikens D, Bruckner-Tuderman L, Uitto J, Wiche G, Schroder R. Severe mucous membrane involvement in epidermolysis bullosa simplex with muscular dystrophy due to a novel plectin gene mutation. Eur J Pediatr. 2004 Apr;163(4-5):218-22. Epub 2004 Feb 13.

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

2<sup>nd</sup> Annual Meeting of International society of stem cell research from June 10-13<sup>th</sup> 2004 Boston, MA

## **HONORS/SPECIAL RECOGNITIONS:**

Dermatology Foundation Research Career Development Grant Award

## **PROFESSIONAL ORGANIZATIONS:**

American Society of Cell Biology (ASCB)

International Society of Stem Cell Research (ISSCR).

## JANET SAWICKI, PH.D.

Associate Professor, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

- Morris, R.J., Liu, Y., Marles, L., Yang, Z., Trempus, C., Li, S., Lin, J.S., Sawicki, J.A., Costarelis, G. Capturing and profiling adult hair follicle stem cells. Nat. Biotechnol. 22:411-417. 2004.
- Anderson, D.G., Peng, W., Akinc, A., Houssain, N., Kohn, A., Padera, R., Langer, R., and Sawicki, J.A. A polymer approach to suicide gene therapy for cancer. Proc. Natl. Acad. Sci. USA. In press.
- Sawicki, J.A. and Peng, W. Prostate tumor regression following adenoviral delivery of Flp recombinase-regulated diphtheria toxin expression. Mol. Ther. 9: S228.
- Peng, W., Anderson, D.G., Langer, R.S., and Sawicki, J.A. Nanoparticle-delivered suicide gene results in prostate tumor regression. Abstract #1188. Amer. Assoc. Cancer Res. Annual Meeting, March, 2004.

# **PROFESSIONAL ORGANIZATIONS**:

American Association for the Advancement of Science American Society of Gene Therapy American Association for Cancer Research International Society of Differentiation

#### DANIEL SHURMAN MD

Post-Doctoral Research Fellow, Department of Dermtology and Cutaneous Biology

## **PUBLICATIONS:**

Topaz O, Shurman DL, Bergman R, Indelman M, Ratajczak P, Mizrachi M, Khamaysi Z, Behar D, Petronius D, Friedman V, Zelikovic I, Raimer S, Metzker A, Richard G, Sprecher E. Mutations in GALNT3, encoding a protein involved in O-linked glycosylation, cause familial tumoral calcinosis. Nat Genet. 2004 Jun; 36(6):579-81.

Shurman D, Reich HL, James WD. Lichen planus confined to a radiation field: the "isoradiotopic" response. J Am Acad Dermatol. 2004 Mar; 50(3):482-3.

Shurman D.L., Glazewski L., Zieske J., Richard G. Calcium-induced changes in connexin expression and function in the corneal epithelium. Abstract at Society of Investigative Dermatology Meeting 2004

Shurman D.L., Glazewski L., Zieske J., Richard G Connexin expression in the human corneal epithelium in vivo and in vitro. ARVO (association of research for vision and ophthalmology) 2004 meeting. Abstract and Paper presentation.

## PARTICIPATED IN SCIENTIFIC MEETINGS AND PRESENTATIONS

Participated in 2004 Annual ARVO and SID Meetings. Paper presentation at ARVO 4/2004- Shurman D.L., Glazewski L., Zieske J., Richard G Connexin expression in the human corneal epithelium in vivo and in vitro.

#### **HONORS:**

Recipient of Fight For Sight Post-Doctoral Fellowship grant 2004. Recipient of National Eye Institute travel grant 2004.

#### PROFESSIONAL ORGANIZATIONS:

Member of the Society of Investigative Dermatology and Association of Research in Vision and Ophthalmology.

#### KENNETH WASSERMAN, M.D.

Adjunct Instructor, Department of Dermatology and Cutaneous Biology

## **APPOINTMENTS:**

Delegate to the Pennsylvania Medical Society Representing Philadelphia, 2004.

Chairman of the American Academy of Dermatology's Major League Baseball Skin Cancer Program: Play Smart When It Comes To The Sun. 2000 - present.

Chairman of the American Academy of Dermatology Task Force for Skin Cancer Screening for Major League Baseball. July 1998 - 2000.

Advisory Board Representative to the American Academy of Dermatology representing the Philadelphia Dermatologic Society. 1992 - present.

Executive Committee-Pennsylvania Academy of Dermatology. 1994 - present.

Co-Chairman, Economics Committee-Pennsylvania Academy of Dermatology. 1995 - present.

Member, Standards of Care Committee-Pennsylvania Academy of Dermatology. 2000 - present.

#### **MEDIA:**

WFAN-AM [IND] "Bob Salter Show" (aud. 290,000) - An interview with Ken Wasserman, M.D., about skin cancer and the *Play Smart When It Comes to the Sun* program aired May 23, 2004.

Comcast [Cable] "News" (comb. aud. 195,000) - An interview with Ken Wasserman, M.D., about sun safety, skin cancer and the *Play Smart When It Comes to the Sun* program aired at 7 p.m. and 10 p.m., May 3, 2004.

KYW-AM [CBS] "News" (aud. 78,600) - An interview with Ken Wasserman, M.D., about sun safety, skin cancer and the *Play Smart When It Comes to the Sun* program aired at various times April 30, 2004.

WSTW-FM/WDEL-AM [ABC] "Lifestyle 1150 AM" (comb. aud. 15,000) - An interview with Ken Wasserman, M.D., about sun safety, skin cancer and the *Play Smart When It Comes to the Sun* program aired at various times, May 1 and May 2, 2004.

## KENNETH WASSERMAN, M.D.

Adjunct Instructor, Department of Dermatology and Cutaneous Biology (continued)

WJJZ-FM [IND] "Philly Focus" (aud. 37, 900) - An interview with Ken Wasserman, M.D., about sun safety, skin cancer and the *Play Smart When It Comes to the Sun* program aired May 30, 2004.

WHYY-TV [PBS] "Delaware Tonight" (aud. 17, 000) - A live interview with Ken Wasserman, M.D., about skin cancer and the *Play Smart When It Comes to the Sun* program aired June 8, 2004.

WUSL-FM [IND] "Philadelphia Focus" (aud. 46, 500) - An interview with Ken Wasserman, M.D., about sun safety, skin cancer and the *Play Smart When It Comes to the Sun* program aired June 13, 2004.

## **MEMBERSHIP IN PROFESSIONAL ORGANIZATIONS:**

American Academy of Dermatology American Medical Association Pennsylvania Medical Society Pennsylvania Academy of Dermatology Philadelphia Dermatologic Society Philadelphia County Medical Society

## **MEETINGS ATTENDED:**

Monthly Philadelphia Dermatological Society Meetings American Academy of Dermatology, Summer Session, New York, July 2004 American Academy of Dermatology, Washington, DC, February 2004 Fall Clinical Dermatology Conference, Las Vegas, NV, October 2003 Pennsylvania Academy of Dermatology, Hershey, PA, October 2003

#### **GUY F. WEBSTER, MD, PHD**

Professor and Clinical Vice Chair, Department of Dermatology and Cutaneous Biology; Professor of Medicine, Division of Clinical Pharmacology (secondary appointment); Director, Center for Cutaneous Pharmacology, Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Reza F. Ghohestani, M.D., Ph.D.; Sherry Rotunda, M.D.; Billy Hudson, Ph.D.; William J. Gaughan, M.D.; John Farber, M.D.; Guy Webster, M.D., Ph.D.; Jouni Uitto M.D., Ph.D.Brief Report: Crescentic Glomerulonephritis and Sub-epidermal Blisters in a Patient with the Predominant IgA Class Autoantibodies to α5 and α6 Chains of Type IV Collagen. Lab. Invest 83:605-611, 2003

MTS Lin, R Gohestani, GF Webster, G. Murphy, Histologic and molecular analysis of epidermolysis bullosa acquisita in an African-American Patient. (Manuscript submitted)

Webster, MD, PhD, Topical Retinoid Therapy in Inflammatory Acne: A Randomized, Masked, Vehicle-Controlled Photographic Assessment. J.A.A.D. in press

AJ Park, GF Webster, RB Penne, IM Raber, Porphyria cutanea tarda presenting as cicatricial conjunctivitis. Am. J. Ophth. 134:619-621, 2002

GF Webster, Laser Treatment for Acne, The Lancet, 362:1342, 2003.

#### **CHAPTERS:**

G.F. Webster, Rosacea, in Dermatology, J Bolognia, J. Jorizzo, R. Rapini eds. Elsevier, London, 2003.

#### **INVITED PRESENTATIONS:**

Moderator, Acne Forum, Summer AAD Meeting, Chicago, 2003
Topical Acne Therapy, Acne Symposium, Summer AAD Meeting, Chicago, 2003
Psoriasis Therapy, Update on Biologicals in Psoriasis, Univ. Pa., Phila, Pa, 2003
Bereston Lectureship, Univ. Maryland, Baltimore, MD, 2003
Rosacea Therapy, Fall Clinical Dermatology Conf., Las Vegas, NV, 2003
Visiting Professor, SUNY Downstate Medical Center, NY, NY 2003
Anti-Inflammatory Activity of antibiotics, Hawaii Derm, 2004
Skin disease in HIV, Caribbean Dermatology, Dorado, PR 2004
Anti-inflammatory antibiotics and rosacea, Caribbean Dermatology, Dorado, PR, 2004
Anti-inflammatory antibiotics, Dallas Dermatological Soc., Dallas, TX, 2004
Visiting Professor, University of Chicago Dept Dermatology, Chicago, IL, 2004
Antibiotic Therapy in Acne, Valley of the Sun Conference, Phoenix, AZ 2004
Visiting Professor, Univ. California, Davis, Sacramento, CA 2004

#### JOHN K. WILDEMORE IV, MD

Resident (PGY-3), Department of Dermatology and Cutaneous Biology

## **PUBLICATIONS:**

Wildemore JK, Lee JB, Humphreys TR. Mohs Micrographic Surgery and Malignant Eccrine Tumors. Accepted for Publication, June 2004 –Dermatologic Surgery Journal

Wildemore JK, Junkins-Hopkins JM, and James WD. Evaluation of the histologic characteristics of patch test confirmed allergic contact dermatitis. Journal of the American Academy of Dermatology. 2003 Aug; 49(2): 243-8.

# PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS:

October 2003: Mohs Micrographic Surgery and Malignant Eccrine Tumors American Society for Dermatologic Surgery Combined Annual Meeting, New Orleans, LA

# **PROFESSIONAL ORGANIZATIONS:**

American Medical Association American Academy of Dermatology American Society for Dermatologic Surgery

#### KYONGGEUN YOON, PH.D.

Professor of Dermatology and Cutaneous Biology Professor of Biochemistry and Molecular Pharmacology Director, Cutaneous Gene Therapy Center

## **PUBLICATIONS:**

Fan, W. and **Yoon**, **K**., "*In vivo* Alteration of the Keratin 17 Gene in Hair Follicles by Oligonucleotide-Directed Gene Targeting", Exp. Derm. 12, 832-842, 2003.

Igoucheva, O. and Alexeev, V. **Yoon, K.** "Oligonucleotide-directed mutagenesis and targeted gene correction: A mechanistic point of view", Current Molecular Medicine, 4, 445-463, 2004

Igoucheva, O. and **Yoon, K**. "Gene targeting by oligonucleotides in keratinocytes" in press, *Epidermal Cell Protocols*, ed, Turksen, K. Humana Press, 2004.

**Yoon, K.** "Oligonucleotide-directed gene correction in epidermis", submitted to J. Invest. Derm., in press, 2004

**Yoon, K.**, Alexeev, V. and Igoucheva, O. "Biased gene repair needs unbiased review", Correspondence, Nature Reviews Genetics, 4, 679-689, 2003.

## **GRANT SUPPORT:**

| 1 R21 AR49229-01 (Yoon) NIH/NIAMS Inhibition of Collagen Synthesis by RNA interference                                     | 09/30/       | 02 – 09/29/04<br>100,000 |
|--|--------------|--------------------------|
| P01 CA072765-06 (PI: Gerwirtz, Project 4 co-PI: Yoon)<br>NIH/NCI<br>"Examining oligonucleotide effects on gene expression" | 8/1/03<br>\$ | 3-6/31/08<br>409,111     |
| <b>5 P01 AR38923-17 (Uitto)</b> NIH/NIAMS Molecular Genetics of Skin Basement Membrane Zone                                | 05/15/<br>\$ | 87-03/31/07<br>655,901   |
| Samsung OTT3210 (Yoon) Samsung Inactivation of ICAM-1 Gene Involved in Psoriasis   | 09/01/<br>\$ | 00-<br>100,000           |
| DE-AC05-00OR22725 (Yoon) DOE (Subcontract) Demonstration of Feasibility of High-Throughput Oligonucleotide                 | \$           | 03 – 3/31/04<br>83,769   |

Point Mutagenesis of Mouse Embryonic Stem Cells

## KYONGGEUN YOON, PH.D.

Professor of Dermatology and Cutaneous Biology Professor of Biochemistry and Molecular Pharmacology Director, Cutaneous Gene Therapy Center (continued)

## PARTICIPATION IN SCIENTIFIC/CLINICAL MEETINGS AND PRESENTATIONS.

#### **INVITED LECTURES**

Pachyonichia Congenita Symposium, Utah, February, 2004

## PROFESSIONAL ORGANIZATIONS.

American Society of Biological Chemistry and Molecular Biology American Association of Advanced Science Society of Investigative Dermatology NIH study section, member of the R21/RFA review group NIH study section, ad hoc reviewer for Medical Biochemistry study section Ad hoc reviewer for the DebRA Foundation grants